

**THE DEVELOPMENT OF A THERAPEUTIC APPROACH FOR THE TREATMENT OF  
INDIVIDUALS WITH PRADER-WILLI SYNDROME AND THEIR PRIMARY  
CAREGIVERS**

by

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## **DECLARATION**

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### **THE DEVELOPMENT OF A THERAPEUTIC APPROACH FOR THE TREATMENT OF INDIVIDUALS WITH PRADER-WILLI SYNDROME AND THEIR PRIMARY CAREGIVERS**

I declare that the above dissertation is my own work and that all the sources that I have used or quoted have been indicated and acknowledged by means of complete references.

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SIGNATURE

30 October 2017  
DATE

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## ABSTRACT

Prader-Willi Syndrome (PWS) is a genetic disorder resulting from a mutation of chromosome 15. It can manifest in physiological characteristics, cognitive impairment, behavioural problems, and sometimes also psychiatric disturbances. Taking care of an individual with PWS has a detrimental impact on the primary caregiver and also affects others around them. This considered, the current study aimed to learn more about the experiences and challenges of individuals diagnosed with PWS and their primary caregivers, in Gauteng and North-West Provinces, South Africa. Purposive sampling was used to select five families which then participated in the study. Qualitative research was used to conduct the study. As it was also crucial to generate a comprehensive understanding of participant experiences, collective instrumental case studies were used – making use of participatory action research, ethnography and elements of auto-ethnography. Data were gathered by conducting semi-structured interviews, which were then analysed using thematic analysis. The data were organized around certain topics and common themes which emerged in each case study and the findings were then integrated with the literature which had been extensively reviewed. Based on these experiences and challenges, interventions were suggested that addressed the challenges and needs of the PWS individuals, their caregivers and families, and those around them (including school teachers). The main findings confirmed that not all individuals diagnosed with PWS manifest all the physiological characteristics, psychiatric disturbances and behavioural problems which have been documented in the literature. Furthermore, the symptoms vary in severity from one individual to the next. Cognitive impairment was, however, common to all individuals in the study. The findings also suggest that having a child diagnosed with PWS has a significantly negative impact on the primary caregiver, and taking care of PWS children is emotionally overwhelming and time-consuming. The use of a client-centred approach, implementing behaviour therapy techniques and doing psycho-education, all proved to be effective in managing some of these behaviours displayed by the individual patients and the challenges experienced by primary caregivers.

**Keywords:** Prader-Willi Syndrome, primary caregiver, behaviour characteristics, cognitive impairment, psychological insights, qualitative research, purposive sampling, thematic analysis, therapeutic interventions, South Africa.

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# CHAPTER 1

## INTRODUCTION

### 1.1 Background information

I am a clinical psychologist working at Dr. George Mukhari Academic Hospital. During the course of my duties I received a referral from the paediatrics clinic at the same institution, requesting psychological intervention for Stephanie, a child diagnosed with Prader-Willi Syndrome (PWS). Stephanie was accompanied by her mother Anna (pseudonyms used). Stephanie presented as a friendly child and was able to answer questions posed to her, and respected me in my role. She appeared short in stature and was slightly obese.

As I was conducting the clinical interview with the mother, Stephanie was interrupting and argumentatively disagreeing with her mother. She did not focus much on the content of what the mother was presenting but rather on putting her point across. I immediately questioned her cognitive functioning. The mother then started explaining the condition (PWS) and provided me with reading material. I was not able to immediately relate, but was listening attentively to every detail and that sparked great interest in me.

My medical psychology knowledge was put to the test. I had no knowledge of this particular medical condition. I was honest with Anna regarding the fact that I had no knowledge of the condition and how it was managed but I was going to focus on treating the presented behavioural problems, as this was my area of expertise. I later consulted with some of my colleagues in order to gather more information on the condition, but none of them could assist me. I went to the library to review the literature on PWS, but I could not find any helpful literature. Although internet searches provided me with adequate information regarding the physical aspects of the condition, they only provided limited information about its psychological aspects.

My quest to understand more about PWS began. I made contact with the Prader-Willi Syndrome Association of South Africa, and Dr. Honey sent me reading material. Her willingness to assist also encouraged me to do a doctorate on PWS.

Prader-Willi Syndrome (PWS), also known as Prader-Labhart-Willi Syndrome, is not widely known in South Africa amongst health-care professionals. Therefore, this chapter will include information on the physical condition and associated behaviours, in order to highlight the challenges faced by the primary caregiver and the individual diagnosed with PWS, from a medical perspective.

The present study was undertaken as there is inadequate literature on the psychological impacts of PWS on individuals diagnosed with it, and also their primary caregivers. I also noted that research on effective treatment methods had been less extensive although various behavioural techniques have been found to be the most effective. This highlighted the need for more treatment methods that could improve the day-to-day life of people with PWS and their caregivers.

Based on the above information, research began on a thesis entitled:

“The development of a therapeutic approach for the treatment of individuals with Prader-Willi Syndrome and their primary caregivers”.

## **1.2 What is Prader-Willi Syndrome?**

Prader-Willi Syndrome was first described in 1958 as a neurodevelopmental genetic disorder which results from a genetic mutation involving chromosome 15. It affects multiple areas in the body of the individual, which leads to dysmorphic features, poor muscle tone (hypotonia), feeding difficulties with sucking deficit and anorexia in the neonatal period, under-developed sex organs (hypogonadism), problems with satiety (hyperphagia), dolichocephaly (a condition where the head is longer relative to its width), cognitive impairment and behavioural and sometimes psychiatric disturbances which affect early development and long term functioning of individuals with PWS. PWS affects both genders equally, all races, all socio-economic strata and occurs in people across all geographic areas (Bar et al., 2017; Hurren & Flack, 2016; McCandless et al., 2011; Mohapatra & Panada, 2016).

PWS is considered to be a spectrum disorder, meaning that not all symptoms occur in everyone affected and may range from mild to severe. The estimated incidence of PWS is from 1 in 15000 to 1 in 25000 live births worldwide (Ho & Dimitropoulos,

2010). PWS is estimated to have affected 400000 people worldwide (Hurren & Flack, 2016).

### **1.3 Genetics and genomics of Prader-Willi Syndrome**

Genetics is a study of heredity, and genomics is defined as a study of genes and their functions. The main difference between genetics and genomics is that genetics scrutinizes the functioning and composition of the single gene whereas genomics addresses all genes and their relationships, in order to identify their combined influence on the growth and development of the organism (World Health Organization, 2002).

Human beings inherit two copies of a gene, one from each parent, and both copies actively shape how we develop. However, if one of those copies is “turned off” and the remaining working gene is defective or severely mutated, then the person may be debilitated. The process where one parent’s contributing gene is “turned off” is called genetic imprinting, and occurs during the formation of an egg or sperm cell. Imprinting means that only one copy of the gene is expressed, while the other is silenced. It is usually the maternal copy that is imprinted, while the mutated paternal copy is not functional. This means while most people have a single working copy of these genes, people with PWS have a non-working copy and a silenced copy. Part of the mechanism that turns off the parent’s gene, is methylation of the parent’s deoxyribonucleic acid (DNA). The genes on chromosome 15 are deleted or unexpressed on the paternal chromosome. Therefore, chromosome 15 is “turned off”, and this leads to the symptoms of PWS (Adams, 2008). Sometimes PWS occurs when a person has two copies of chromosome 15 inherited from the mother, instead of one copy from each parent. This phenomenon is called maternal UniParental Disomy (mUPD) (McCandless et al., 2011).

### **1.4 How Prader-Willi Syndrome is diagnosed**

Different tests are available in South Africa for confirming the PWS diagnosis. Testing for PWS begins with DNA methylation analysis as it is the most efficient and cost-effective screening test. If the methylation analysis is consistent with PWS, fluorescence *in situ* hybridization (FISH) is used to detect a deletion on chromosome 15, and if it reveals no evidence of deletion the next step is to obtain blood from the

parents and the child to evaluate for mUPD. If biparental inheritance is discovered in the face of abnormal methylation and normal FISH results, then by a process of elimination, the cause is assumed to be an imprinting defect (McCandless et al., 2011).

Christianson, Viljoen, Winship, De la Rey, and Van Rensburg (1998) investigated whether PWS was over diagnosed, and confirmed it in only 35% of South African patients who have had a PWS diagnosis, confirming that this condition is over diagnosed and that the clinical diagnosis is difficult. The clinical diagnosis is considered difficult because many features are subtle or non-specific, and others change with age. The authors did suggest that the incidence of PWS in the black population should be lower than in the Caucasian population in South Africa, because of early mortality secondary to feeding problems and failure to thrive.

According to Whittington and Holland (2010) compared to 20 years ago there is now a generation of children who have been diagnosed with PWS within days or weeks after birth. Bar et al. (2017) found that the mean age at diagnosis was 18 days. These children have access to food in a managed environment, and, as a result, gross obesity has been avoided, and with growth hormone supplementation many have normal growth trajectories and a final height compatible with parental height. With the establishment of a national PWS Association and the availability of information through the internet, parents are now much more knowledgeable (Whittington & Holland, 2010).

#### **1.4.1 Diagnosis during the neonatal period**

McCandless et al. (2011) and Hurren and Flack (2016) state that PWS should be considered in any infant with significant poor muscle tone (hypotonia), accompanied by poor feeding and reduced spontaneous arousal for feeding. Significant neonatal hypotonia is present in essentially all children for whom molecular testing confirms the diagnosis of PWS. Therefore, this history should be actively sought during the evaluation of older children.

### **1.4.2 Diagnosis during childhood**

Under-developed sex organs such as undescended testes, a small phallus, or small clitoris (hypogonadism) is also an indicator. So is increased appetite and excessive weight gain from 2 to 3 years of age. In older children, the diagnosis should be considered when there is impaired satiety for food, especially with rapid weight-gain. Poor linear growth, especially in the presence of excessive caloric intake, should also raise suspicion for PWS (McCandless et al., 2011).

Developmental delays, speech-articulation defects, and a characteristic physical appearance should all raise the index of suspicion. Individuals with PWS are typically short, obese, possess small hands and feet, and have dysmorphic features like a narrow bifrontal diameter, full cheeks, almond-shaped eyes, dental caries and small mouth (with downturned corners of a dry mouth with sticky saliva and thin upper lip) (Butler, Manzardo, Heinemann, Locker, & Locker, 2016; Ho & Dimitropoulos, 2010; Hurren & Flack, 2016; Mohapatra & Panada, 2016; Sinnema et al., 2011).

Some children with PWS may have sleep disorders, including disruptions of the normal sleep cycle, excessive day time sleepiness, sleep apnea (a condition in which breathing pauses during sleep), temperature instability and/or insensitivity, and a high pain threshold (Whittington & Holland, 2010).

## **1.5 Characteristics of individuals with Prader-Willi Syndrome**

### **1.5.1 Hypotonia**

In individuals with PWS, hypotonia is prenatal in onset, and usually manifests as decreased foetal movement, abnormal foetal position at delivery, and there may be an increased need for assisted delivery or caesarean section. In infancy, there is decreased movement and lethargy with decreased spontaneous arousal, weak crying, and poor reflexes, including poor sucking that leads to early feeding difficulties and poor weight gain. Assisted feeding through a feeding tube and/or special nipples with increased feeding times are necessary, usually for weeks to months. Mild to moderate hypotonia persists throughout life. Hypotonia is a universal characteristic of this syndrome, and all newborns with unexplained, persistent hypotonia should be tested for PWS (Cassidy & Driscoll, 2009; Cassidy, Schwartz, Miller, & Driscoll, 2012; Ho & Dimitropoulos, 2010).



### **1.5.2 Hypogonadism**

Hypogonadism occurs when the body's sex organs produce little or no hormones. In men, these organs are the testes and in women the ovaries. In individuals with PWS, hypogonadism has been observed in both sexes and manifests as genital hypoplasia (under-developed genitals) throughout life, incomplete pubertal development, and infertility in the vast majority. In males, the penis may be small, but most characteristic is a hypoplastic (under-developed) scrotum that is small, poorly rugated, and poorly pigmented. In females, the labia and clitoris are generally hypoplastic (under-developed). In males, when pubertal changes would normally be expected, there is poor voice change and also scanty beard and body hair. In both sexes, little or nothing is known about sexual activity which is believed to be lacking, and most affected individuals are presumed to be infertile. In very rare instances, affected females have had babies (Cassidy & Driscoll, 2009; Cassidy et al., 2012).

### **1.5.3 Developmental and cognitive delays.**

Cassidy and Driscoll (2009) stated there are delayed milestones, including gross motor and language delays. Early milestones are reached on average at double the normal age for example, sitting at 12 months, walking at 24 months, and speaking words at 2 years.

Mental retardation has also been considered an integral part of PWS (Whitman, 1995). Cognitive disability is evident when individuals start school. Mild to moderate cognitive impairment is a common characteristic of PWS, with about 40% having borderline mental retardation or low-normal intelligence, and approximately 20% have moderate retardation (Cassidy & Driscoll, 2009).

Regardless of the IQ score, most individuals with PWS have multiple severe learning disabilities and poor academic performance. Some people with the syndrome, however, will test solidly in the normal range on standardized IQ tests. However, they still appear to have limitations compared to IQ-matched peers in the general population, and function like people with mild mental retardation. Abstract concepts and concepts of time, in particular, present difficulties for people with PWS (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

According to Whittington and Holland (2017) many researchers report numerical skills to be one of the most problematic areas of cognitive functioning for people with PWS. In the study conducted in the United Kingdom, participants could not do any calculations, read, spell and expressive language was observed to be poorer than receptive language. Whittington and Holland (2017) also stated that many parents reported that their PWS children had difficulty understanding what is said to them and often “get hold of the wrong end of the stick”. This was attributed to the fact that PWS individuals have a tendency to interpret everything literally, and have a concrete way of thinking.

By school age, in addition to intellectual disabilities, social difficulties become more apparent (Whittington & Holland, 2010). They exhibit higher overall behaviour disturbances compared to individuals with intellectual disability of unknown cause (Ho & Dimitropoulos, 2010). Most adults require sheltered residential and employment settings because of a combination of cognitive, behavioural and food-seeking characteristics (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

Because social cognition may also be impaired, most people with PWS have difficulties relating to their peer groups, and often prefer to be with older or younger groups. Some of the social difficulties are similar to those seen in people with autism spectrum disorder (Whittington & Holland, 2010; Whittington & Holland, 2017). People with PWS may withdraw into solitary activities, for example, doing word puzzles and jigsaw puzzles, rather than undertaking activities with their peers (Whittington & Holland, 2010). It has been evident that individuals with PWS have exceptional skills with jigsaw puzzles, they use the shape of the pieces rather than the picture or colour as their main strategy, and this advantage disappears when pieces are cut by a straight line rather than traditional jigsaw shapes (Whittington & Holland, 2017).

Early childhood is often characterized by rigidity, particularly related to daily routines, and there is long-term persistence of temper tantrums, stubbornness, and manipulative and oppositional defiant behaviours typical of the normally developing two-year-old. It is later observed that the children have perseverant speech and

obsessive-compulsive behaviours (repeated organizing, writing, collecting, need to finish one thing before moving to the next), and particularly skin picking becomes prominent (Ho & Dimitropoulos, 2010; McCandless et al., 2011). Clarke, Boer, Chung, Sturney and Webb (1996) suggested that temper tantrums, self-injury, impulsiveness, lability of mood, inactivity and repetitive speech are maintained as characteristic behaviours in PWS adult life.

#### **1.5.4 Skin picking**

According to Gourash and Foster (2005) the skin picking behaviour of PWS has a wide range of severity from patient to patient and sometimes in the same patient over time. Some patients have occasional minor skin picking while others maintain large open wounds. Skin picking can sometimes be severe enough to require hospitalization. Skin picking is defined as an activity that goes on when the patient is calm, and does not appear to be a result of expressing emotional distress. The phenomenon has been related to boredom and anxiety, but objective evidence for this is difficult to establish (Whittington & Holland, 2010).

No specific intervention has been effective for skin picking. The behaviour is often extinguished if healing of the wound is achieved. Behavioural interventions have however been effective in some cases. Because skin picking behaviour occurs intermittently and secretively, behavioural interventions targeted at the activity itself are difficult to implement. A basic principle is that no attention, positive or negative, should be paid to the behaviour itself other than to require the patient to adhere to good manners and good hygiene (Whittington & Holland, 2010).

#### **1.5.5 Food-related behavioural problems**

According to King (2008) two separate and distinct eating disorders are noted: initial feeding difficulties and failure to thrive, and later, overeating. In the second phase the disorder ensues with hyperphagia (excessive appetite for food), which King (2008) stated seemingly begins in late childhood. The hyperphagia is hypothalamic in origin, and results in a lack of satiety.

Honey (2010) states that the changing eating pattern becomes evident as early as two years of age. It causes major concerns such as the over-eating and severe

obesity, which then become a central feature of the syndrome. The reason for these appears to be a failure of the satiety response following food intake. Honey (2010) further highlighted that the vast majority of individuals with PWS eat continuously and show no slowing down of their eating behaviour when plenty of food is available.

Visual analogue scales used to assess feelings of hunger and fullness before, during and after food intake, showed that people with PWS did eventually satiate but only after eating three times more calories than a control group. The feeling of hunger also returned shortly after food was no longer available. It seems that the normal feedback mechanism, which causes the loss of feelings of hunger and increased feelings of fullness leading to cessation of feeding, is faulty (Honey, 2010).

Subsequently, children with PWS may develop a wide range of food-related behaviours, including food seeking (which is common), hoarding, foraging for food, eating of unappealing food items, and stealing of food or money to buy food, eating non-food items (e.g. animal chow, spoiled food, decorative items that look like food, searching in garbage cans), and even running away from home to search for food over a wider area – which is potentially quite dangerous and difficult to manage. Typical of the adolescent years, the teenager with PWS is often overly confident of his or her ability to handle risks and dangerous situations (Honey, 2010; McCandless et al., 2011).

If food intake is not controlled externally, obesity results from the above behaviours, which is combined with a low metabolic rate and decreased activity level. Complications of obesity are the major causes of morbidity and mortality, for example, cardio-respiratory insufficiency, obstructive sleep apnea, thrombophlebitis (vein inflammation), and chronic leg oedema (swelling). Up to 25% of obese adults have type II diabetes mellitus, with a mean age of onset of 20 years (Cassidy & Driscoll, 2009). With careful weight control, people with PWS can remain healthy well into older adult life, and some have been known to live into their seventh decade (McCandless et al., 2011; Shriver, 2004). According to an article from the International Prader-Willi Syndrome Organization (IPWSO, 2013) psychological stress of social stigmatization imposed on obese children may be just as damaging as the medical morbidities.

### **1.5.6 Behavioural problems**

Almost everyone with PWS presents with behaviour which can bring challenges to themselves or those around them. The behaviour challenges may vary from mild to severe (Prader-Willi Syndrome Association United Kingdom [PWSA UK], n.d.). It has been observed that characteristic behavioural patterns begin in early childhood in 70-90% of affected individuals and are typified by temper tantrums; food seeking; stubbornness; resistance to change; argumentativeness; controlling, attention-seeking and manipulative behaviour; perseveration, skin picking, lying and blame shifting, compulsive behaviours, and difficulty with change in routine (Cassidy & Driscoll, 2009; PWSA UK, n.d.). Many of the behavioural characteristics suggest the autism spectrum disorder, for example, attention deficit/hyperactivity symptoms, rigidly sticking to routine and repetitive behaviours are common and of early onset. The severity of behavioural problems increases with age and body mass index, and then diminishes in older adults (Cassidy & Driscoll, 2009; PWSA UK, n.d.).

Several factors can worsen these behaviours, such as but not limited to: major and minor changes in daily routine; changes in diet or meal routine; conflicts of opinion among others in a close relationship to the person with PWS; unexpected happenings or upsets; and major life stressors such as bullying, teasing, bereavement, moving residence, changing schools, tiredness and the person being unable to get their way (PWSA UK, n.d.).

The attitudes of other people can also trigger the above stated behaviours. These include aggressive attitudes and tone of voice, confrontational approach, being overly sympathetic, displaying dislike of the person, not talking to the person, talking to them too much, contradicting information from parents and saying “don’t” as a means of trying to stop the individual with PWS from doing something (PWSA UK, n.d.).

PWSA UK (n.d.) further indicates that behavioural difficulties in individuals with PWS are influenced by different factors, which may include cultural and social influences and parental or carer attitudes. Genetic factors also contribute to behavioural difficulties, and are associated with some dysfunction in the hypothalamus. It is

therefore important to bear in mind that certain aspects of PWS behaviour derive directly from physical disturbances in the brain.

According to Schwartz et al. (2016) factor analysis suggest that the behaviour could be grouped into three categories: a) eating disorder, lying and stealing; b) repetitive and ritualistic behaviours and temper outbursts; and c) skin picking and mood disorders. Each of these three groups may have different causal mechanisms. For example, impaired set-shifting ability and different patterns of brain activation have been shown to be associated with increased temper outbursts, when routines are disrupted, and with repetitive and ritualistic behaviour.

Implementing the following will likely have a positive effect on the individual with PWS: firm, but loving and caring attitudes; having a sense of humour; presenting clear guidelines and boundaries and sticking to them; maintaining positive social contacts with the person; praise and stressing the positive aspects of the person with PWS; and saying “do” as a way of showing the person how to do something correctly (PWSA UK, n.d.).

Despite the behavioural problems it is important to note that people with PWS have positive characteristics. They are known to be friendly, sociable, kind and caring, and many have a wonderful sense of humour (PWSA UK, n.d.).

Whittington and Holland (2010) found that the families of people with PWS are subject to more stress than those of people with imprinting defect (ID) mixed aetiologies, and 70% of mothers have high levels of stress needing psychological counselling. Parents are divided on which particular phenotypic characteristic is most stressful for them, but most cite the eating behaviour or obsessive-compulsive behaviour/s depending on which is predominant in their child’s behaviour. Closely related to these two characteristics is the problem of temper outbursts which most often occur when expectations are not met such as expectations about food and routine, but also of concern is extreme hoarding behaviour. Worries about the affected child’s health and well-being also cause stress for parents, and in the case of severe skin picking and psychiatric illness may be exacerbated by social stigma.

### **1.5.7 Psychiatric disturbances**

Cassidy and Driscoll (2009) reported that psychosis in individuals with PWS is evident by young adulthood in at least 5-10% affected individuals. Sinnema et al. (2011) also confirmed that the mean age of onset of the first psychiatric episode was 21 years and 9 months. They established that family history had little influence in the development of psychiatric illness in adults with PWS. In addition to the genetic vulnerability, psychosocial stressors are known to be triggers for the development of the first psychotic episodes. Stressors for their participants included changes in living circumstances, graduation from school and starting work, or death of a family member (Sinnema et al., 2011).

### **1.5.8 Personality traits.**

According to Honey (2010) a Belgian study differentiated two distinctive personality patterns: an “active or extroverted” group and a “passive and introverted” group. The children with extroverted tendencies showed poor social skills and were demanding, attention-seeking, persistent, easily frustrated and rigid, and with frequent temper tantrums. They were able to express their feelings and needs, but their distinction between fantasy and reality was poor.

In their early childhood, children with extroverted tendencies showed features of autism like impaired eye contact, inappropriate physical contact, and a lack of varied, spontaneous make-believe or social play (Honey, 2010). Also observed was the need for control over rituals which often evolved into obsession. Anxiety levels were high, with frequent panic attacks and temper tantrums. Affected children liked to have control in different situations. Speech problems included word-finding difficulties, speech dysfluency, and endless verbal perseveration mainly about subjects like food. Problems became more severe under stressful conditions (Honey, 2010).

The children with introverted tendencies showed social withdrawal with poor peer relations, did not want to participate in group or team games, and liked to be mothered by their peers. Such children avoided physical activities and preferred quiet games like drawing, reading, and listening to storytelling. Temper tantrums occurred less frequently.

They exhibited behavioural problems which were internalized. Bullying by peers and exclusion from games were common, and under stressful conditions these children were passive and withdrawn and their internalized feelings resulted in anxiety and depressive symptoms. Temper tantrums, rituals, obsessive behaviour and the need for control were less extreme. Periods with emotional lability, sudden crying, unhappiness, and general dissatisfaction were alternated with cheerfulness and general dissatisfaction (Honey, 2010).

In adolescence, both groups started to manifest psychiatric symptoms which seemed to fluctuate spontaneously over periods of several hours to months. Children had periods with good control of diet, weight loss, a stable emotional life, general feelings of happiness alternated with poor weight control, weight gain, and psychiatric symptoms. Children with extroverted tendencies showed more agitation, refusal of human contact, and even had hallucinations with mild anxiety whereas children with introverted tendencies had mood swings, low frustration tolerance, emotional instability and irritability, and also sleeping and eating disturbances. Children with introverted tendencies showed very high anxiety levels. Both groups were treated with behaviour modification and medication (Honey, 2010).

## **1.6 Mortality**

Several reports and surveys of deaths in persons/patients with PWS have found that obesity-related, cardiovascular and respiratory disorders were the most frequent causes of death in both children and adults. Specific concerns have been raised about eating related fatalities, including choking on gorged food and gastric necrosis and rupture following bingeing, particularly in slim but previously obese individuals (Cassidy & Driscoll, 2009).

Butler et al. (2016) found that respiratory failure was the most common cause of death in adults and children with PWS whilst cardiac disease and failure with pulmonary thromboembolism were commonly found in adulthood in combination with obesity-related morbidity. Cardiopulmonary and body mass index (BMI)-related mortality factors predominated among females while males were more likely to experience accidents, choking, and infection at a young age. Accidents, pulmonary



aspiration, sepsis and choking were the most common cause of death in PWS children.

## **1.7 Treatment and management of individuals with Prader-Willi Syndrome**

### **1.7.1 Multidisciplinary approach**

Children with PWS should receive early intervention (including physical, occupational, and speech therapies) and individualized, appropriate education. This ideally includes social skills training. Optimally, a one-to-one aide in the classroom will help the child focus on learning and diminish behavioural disturbances (Cassidy & Driscoll, 2009).

### **1.7.2 Management of hypotonia**

During management of hypotonia in infancy, the major focus should be on assuring adequate nutrition and growth. An infant with PWS rarely wakes up to feed, and therefore a regular feeding schedule should be established. Early infant stimulation programmes are strongly recommended to ensure adequate interactions and to optimize strength and milestone achievement. Encouragement of physical activity is important for strength and agility at all ages (Cassidy & Driscoll, 2009).

### **1.7.3 Growth Hormone Therapy (GHT)**

The benefits of growth hormone therapy in infants, children and adults with PWS have been well-demonstrated in multiple well-designed and well-controlled studies. Treatment can begin as early as two to three months of age. It is important that parents be thoroughly informed about the potential benefits and potential for undesired effects. For example, there have been several deaths in children as young as three years with PWS within six months of initiating GHT. The role of GHT in these deaths, if any, is not known (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Whittington & Holland, 2010).

There is evidence from controlled clinical trials that GHT in children improves stature, muscle mass, facial appearance and bone density, head circumference, height, body mass index, body composition (with improvement of lean muscle mass and delay of fat tissue accumulation), body proportions, acquisition of gross motor skills, language acquisition, and cognitive scores. Growth hormone treated children breathe better

due to stronger respiratory muscles and an improved response to the build-up of carbon dioxide (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Whittington & Holland, 2010).

#### **1.7.4 Managing food-related behaviour**

IPWSO (2013) found that no appetite suppressant has worked consistently for people with PWS. Most require an extremely low-calorie diet all their lives and their environment must be designed so that they have very limited access to food. Strategies to limit access to food include consistent limit setting and close supervision at all ages, including locking of cabinets and the refrigerator at home, limiting exposure that makes the child think about food, and avoidance of work environments with available food and these decrease anxiety and conflict (IPWSO, 2013). It is also important to keep dustbins locked and to alert neighbours and shopkeepers to the child's problem especially if stealing, or begging for food or money is a particular difficulty. It should also be ensured that others do not eat in front of the person with PWS (PWSUK, n.d.).

It is imperative to begin good meal management and education at an early age. This includes sticking to a strict schedule for meals and snacks, and also limiting portion sizes. Confidence should be instilled that the next meal will be served on time, by scrupulously maintaining mealtime routines. Relatives and social contacts must be educated to realize that "sneaking" food to the child with PWS is not an appropriate method of demonstrating affection, and, in fact, undermines the child's nutritional regimen and sense of well-being (IPWSO, 2013).

Nutritional counselling for good long-term weight management should begin in early infancy, in order to prevent inappropriate weight gain that would otherwise typically begin from 12 to 36 months of age. Exercise is also a very important factor in weight maintenance, and early establishment of a routine of regular daily physical activity, of at least 30 minutes, is strongly recommended (IPWSO, 2013).

PWSA UK (n.d.) states that the above suggested interventions will not work with everyone, and that some behaviours are reinforced if they result in the individual getting what they want. The individual may "act up" in one environment where his or

her behaviour is “rewarded” by others “giving in” but will not exhibit the behaviour in an environment where others take a firmer stance. Individuals with PWS experience genuine remorse after an outburst, but it does not seem to prevent further such outbursts (PWSA UK, n.d.).

### **1.7.5 Psychiatric medications**

Behavioural problems should be detected early on and treated appropriately with parental education/training (including consistent limit setting), and, if needed, with counselling and/or psychotropic medication (Cassidy & Driscoll, 2009). It is not always possible to correct the genetic abnormality; therefore, the treatment is often aimed at suppressing unwanted symptoms (Mohapatra & Panada, 2016). Serotonin agonists have been the most successful in reducing temper outbursts and in improving compulsivity (different medications are discussed below). Families should be made aware of the signs of psychosis and urged to undertake psychiatric assessments early, if a thought disorder is apparent (Cassidy & Driscoll, 2009).

- ***Antidepressants***

The class of antidepressants most commonly used for PW patients is the selective serotonin reuptake inhibitors (SSRIs). These medications have the advantage of being very easy to give. They are usually given once a day, have few and easily manageable side effects, and are safe when taken as an overdose. For these reasons, they are usually tried first. They are useful, not just for depression, but also for other problems which affect PW patients, such as obsessive-compulsive disorder (OCD), aggression and impulsive behaviour (Boyle, 1997).

- ***Anti-psychotics***

According to Boyle (1997) these are a second class of drugs that is often prescribed for PW patients. Some PW patients have brief psychotic episodes and benefit greatly from antipsychotic medications. The use of these medications is not limited to delusional patients. They are also used for patients with bipolar affective disorder, as well as for those with agitated, aggressive behaviour, and very impulsive behaviour.

- ***Anti-anxiety agents***

This is the third class of medication. Many patients with PWS are rigid and quite anxious and benefit from anti-anxiety medications (Boyle, 1997).

- ***Mood stabilizers***

Boyle (1997) states that the fourth class of medication is the mood stabilizers and they are tried with PW patients who suffer from aggressive or impulsive outbursts. While these medications may help, patients with PWS must also be in a controlled, structured environment, in order to truly benefit from the medication.

- ***Psycho-stimulants***

This is the final class of drugs used for PW patients. It is now recognized that the medications may well be helpful for PW patients, who certainly have attention difficulties (Boyle, 1997).

### **1.7.6 Behaviour modification and management**

#### **1.7.6.1 Behaviour modification**

Following the detailed outline of the challenges, it is clear that the three most outstanding characteristics of PWS are the cognitive impairment, behaviour and food-related problems (especially problems with satiety), which are evidently interrelated. This section offers suggestions for behavioural modifications that have had some success and which hopefully can be used with variations to devise helpful management for all involved. These will be implemented during the research process in order to assess their effectiveness, and will also facilitate the development of the most effective intervention method.

McCandless et al. (2011) state that management of many complex behavioural issues is best accomplished through active partnership of the parents, the primary care provider, and a developmental or behavioural specialist (paediatrician or psychologist). Behavioural management that focuses on rewarding desired behaviours and which ignore, when possible, undesirable behaviours seems to be the most effective.

Early recognition of developing behaviour problems is critical for maximizing the effectiveness of such an approach. Parents should be advised that offering food as a reward, or withholding food as a punishment, is almost always counterproductive and should be avoided. Positive reinforcers are generally not difficult to identify, and reward systems that use small, short-term goals that progress to larger goals are effective (McCandless et al., 2011).

#### **1.7.6.2 Behaviour management**

Since by definition persons with mental retardation have cognitive limitations, behaviour modification treatment methods have superseded traditional psychotherapeutic treatments in these populations. Behaviour procedures are among the most frequently used to treat, manage, and/or control the multiple emotional and behavioural difficulties presented by persons with mental retardation (Whitman, 1995).

These treatment procedures are used either to stop or reduce the frequency, duration and intensity of unwanted behaviours or to increase appropriate behaviours. Fundamental to all these approaches is the assumption that even the most cognitively impaired person eventually learns from the experience of a behaviour-consequence pairing (Whitman, 1995). While these procedures are occasionally successful when used with PWS to increase certain behaviours, they fail badly at eliminating the neurologically-driven inappropriate behaviours. Viewed from a different perspective, these behaviour treatment methods affect learning and change through the use of reactive (often punitive) methods of behaviour change at the expense of proactive, planned behaviour management and behaviour guidance. The sequential deficits noted in persons with PWS suggest that a reactive form of behaviour management will be ineffective (Whitman, 1995).

According to Whitman (1995) behaviour management must focus on preventing the opportunity for expression of such behaviours rather than depending on the development of internal cognitive controls. Behaviour management strategies differ from behaviour modification as follows:

- Management procedures are initiated prior to the occurrence of the problem behaviour.
- Management procedures remove or minimize conditions that instigate problem behaviours.
- Management procedures present or emphasize conditions that increase the likelihood of appropriate behaviour and decrease the opportunity for inappropriate behaviour.
- Management procedures minimize the duration and intensity of problem behaviours following their occurrence. The components of these procedures include:

- ***Environmental structure***

Environmental structure must anticipate the aspects of the neurological drivenness in the person with PWS. Both the physical environment and decision-making environment need to be structured. Since most adults with PWS cannot make wise life decisions for themselves, and do not adhere to decisions once made, external guardians must be responsible for placing and maintaining the PW person in an appropriately protected living and work setting (Whitman, 1995).

Funds should be managed such that no money is available to the PW person for food purchases beyond that allowed in the dietary programme. Some may need to have their telephone privileges monitored as they are known to call food delivery services. The physical living environment must be designed so that food delivery, food access, food preparation and food disposal are managed in a fashion that is neither visible nor accessible to the person with PWS. This usually implies locked cabinets, pantries, refrigerators, and, ideally, totally locked kitchens (Whitman, 1995).

The physical environment must anticipate that PWS persons have the inordinate ability to pilfer other people's food, to take food from machines, or to manipulate others into giving them food. Recreational outings must also anticipate food distribution areas and the outing should be managed in such a way that such areas are avoided (Whitman, 1995).

- ***Operational guidelines***

Rules, regulations and procedures must be spelled out, written down and prominently displayed. When displayed in a clearly visible area, these guidelines serve to alleviate many power struggles and tantrums, by pointing to the posted rules and thereby eliminating the symmetrical struggle between the PW person and a staff or family member. Together with these general guidelines, any individual behaviour goals and the means for achieving them can be posted as can rewards and honours for successes achieved (Whitman, 1995).

- ***Time***

Persons with PWS understand time only from the concept of 'now'. Past time and future time mean little. Thus, visual representations of time can help the person with PWS understand how long it will be until something will happen or they can provide them with a visual tool when waiting for it to be time to do something (Whitman, 1995).

- ***Choice and reward management***

Whitman (1995) report that open-ended options will generally lead to disastrous results for individuals with PWS. If they are given open-ended options they will be unable to make a decision and will get anxious or they make a decision that is inappropriate. Choice management will offer the choice between two or more alternatives but always alternatives with the same end. For example, "do you want to go to the zoo or the ballgame?" will elicit a choice between two options, while "what do you want to do today?" may result in hours of indecision, and ultimately, a tantrum when someone else finally takes the decision (Whitman, 1995).

- ***Consequences***

Consequences of compliance or non-compliance need to be known ahead of time. Reactive consequences usually lead to a power struggle and a tantrum. In addition, consequences must be enforced without fail. Any change leads to a toppling of the whole structure as the person with PWS is inordinately attuned to other peoples' soft spots and can manipulate them, almost at will. The most efficient form of

consequence management is one in which desired behaviour must be earned rather than undesired behaviours punished (Whitman, 1995).

- ***Anticipatory planning***

According to Whitman (1995) good management means always anticipating tomorrow, next week, new situations, changed plans, and the like. For example, a school outing can be a success or failure depending upon the adequacy of the planning. Pre-planned lunches, pre-stated rules, and pre-determined and stated times to be spent in any one place and with a specific person. Clearly delineated authority figures and anticipated rewards and consequences provide the structure for behaviour with clear limits and without ambiguity (Whitman, 1995).

- ***Behavioural rehearsal and rewards for flexibility***

Whitman (1995) state that an inability to shift perspective, cognitive rigidity, and a failure of sequencing abilities that facilitate problem solving render the person with PWS vulnerable when change is demanded. This is a vulnerability that is usually coped with by stubbornness, refusal to act, and, ultimately, a tantrum. Anticipating ahead of time any possible outcomes, and rehearsing these ahead of time, gives the person with PWS a structure and a familiar repertoire to enable coping when change is required. Memory can be jogged with “remember, we practiced how to do this”. A routine for not arguing should be established. For the older child who needs to feel some participation in decision making, a time limit should be set as to when they should stop talking (Whitman, 1995).

- ***Tenuous emotional control***

According to White (1998) any seemingly minor combination of life stressors can contribute to loss of control as evidenced by tantrums and self-injury. Once control is lost, it is typically a period of time before it is regained. This is often followed by feelings of sadness, remorse and guilt. It is necessary to be vigilant to life stressors and to avoid or prepare the person for the change. It is important to read early signs of control loss and to provide support. Once control is lost, a safe area should be provided where they wait it out, and afterwards provide a “talk out” in order to avoid negative consequences (White, 1998).



- ***Peer social interactions***

Social interactions are difficult. While persons with PWS need and value friendships, it may be an emotional strain to be exposed to the unpredictability of others for extended periods. They often lack age-appropriate social skills, and face challenges in issues of fairness and comparing themselves to others which often results in frustration and anger. As a form of support, one-to-one or small groups may be preferable. There must be a time limit on social activities. Routine and structure to activities should be provided (White, 1998).

- ***Stress associated with support to PWS individuals***

The amount of energy required to monitor and support persons with PWS is extraordinary, and family members are especially at risk of developing feelings of stress. As a form of support, respite and shared parenting should be provided for families. Out of home support for periods of time and sibling support should be provided or considered while professionals should practise shared care giving. Strategies of personal emotional equilibrium should be practiced (White, 1998).

### **1.7.7 Managing temper tantrums (PWSAUK, n.d.)**

Temper tantrums should be managed as follows:

- Remove the individual from the situation or remove the audience.
- Ensure their safety and that of others.
- Stay calm, and talk as softly and as little as possible.
- If possible, get someone else to take over if the outburst is directed at you.
- Do not attempt to argue or reason with the individual.
- Do not give in to their demands.
- Try distraction and even humour.
- After an outburst, the cause of the outburst must be identified and possible solutions found.
- Other ways of assisting the individual could include basic anger management techniques like: making use of relaxation tapes, assisting the individual to identify physical feelings associated with emotional states so they can recognise in advance when they might lose control, taking a walk, listening to music, or distracting them.

### **1.7.8 Managing obsessive-compulsive behaviour (PWS, n.d.)**

Obsessive-compulsive behaviour should be managed as follows:

- Provide a range of activities especially social activities where PWS persons will meet new people.
- Suggest that hoarded objects be sold, in order to make money for a special treat.
- Reduce the opportunities to be with the person or thing the PWS person is obsessed with, and support the PWS person to appreciate that they may need to remove themselves physically.
- Have someone else give the person with PWS some attention, in order to divert attention away from the person they are obsessed with.
- Use time spent with the favourite person as a “reward” for good behaviour for not bothering the particular person for the rest of the day but limit the time to around 15-30 minutes.
- If obsessions are severe and interfere with the PWS person’s quality of life, medication from a psychiatrist or similarly qualified person may help.

### **1.7.9 Managing lying and blame-shifting (PWS, n.d.)**

Lying and blame-shifting should be managed as follows:

- Do not agree with the lies, but do not be confrontational as this will escalate the PWS person’s sense of being out of control in relation to the situation.
- Monitor the individual’s activities to ensure that opportunities do not arise which would in fact trigger lying.
- Ensure good communication within the environment so that everyone knows as much as possible about what is happening on a daily basis, and will thus be able to spot more easily when the PWS person is lying.
- Negotiate a contract with a person, so they are clear what would happen if they do lie.
- Ensure that the PWS person knows when they will get into trouble for doing something, and when it may not be their fault. Sometimes they will lie or shift the blame just because they think they may be in trouble.

### **1.8 Research problem and motivation**

During infancy many PWS children display a range of behavioural problems that become more noticeable in adolescence and adulthood, and which interfere mostly with their quality of life. Early diagnosis of PWS is important for effective long-term management, and a multidisciplinary approach is fundamental to improving quality of life, preventing complications, and prolonging life expectancy. A multidisciplinary team consists of neonatologists, paediatricians, endocrinologists, orthopaedic surgeons, psychologists, psychiatrists, physiotherapists and urologists (Mazaheri et al., 2012).

The quality of life for the individual with PWS depends on the ability of caregivers to provide an environment which is structured enough for the PWS individual to minimize temptations like accessibility to food which could contribute to other medical problems like obesity. Maintaining such a structure is however, labour intensive for parents and other caregivers. Failure to maintain an appropriate structure is debilitating for all concerned, especially the individual with PWS (Whitman, 1995). Caring for individuals with PWS requires inordinate understanding and patience. The total involvement of family and carers is essential to ensuring a restricted environment (Mazaheri et al., 2012).

Moss (2009) found that heavy demands are made on parents and caregivers of individuals with PWS which usually leads to negative experiences and feelings. Whittington and Holland (2010) found that families of people with PWS are subject to stress and 70% of the mothers have high levels of stress, which needs psychological counselling. Cassidy and Driscoll (2009) suggested that behavioural problems should be detected early, and treated appropriately with parental education/training (including consistent limit setting), and, if needed, consideration of counselling and/or psychotropic medication. Hence it is important to not only focus on the diagnosed individual but also to provide necessary intervention to the primary caregiver as they are confronted by multiple stressors.

Mazaheri et al. (2012) found that families, mothers and siblings with PWS reported difficulties in family functioning, communication problems, and an increased number of conflicts. The study reaffirmed that PWS affects the entire family system. Mothers

and siblings would benefit from psychosocial support due to the multiple challenges of living with and caring for a child or young adult with PWS. Mazaheri et al. (2012) suggested that interventions should focus on providing psychosocial support for each member of the family, and should encompass all aspects of life such as the emotional, physical, issues of communication, family dynamics, and personal perceptions of disease.

Based on all the above statements and findings it is important to conduct psychological research to build up knowledge on the experiences and challenges of both the individual with PWS and of the primary caregiver. It is also necessary to develop therapeutic interventions relevant to the PWS individual, and the primary caregiver.

### **1.9 Objectives**

Objectives are to:

- Learn about the experiences and challenges of individuals diagnosed with PWS.
- Learn about the experiences and challenges of primary caregivers of individuals with PWS.
- Develop therapeutic methods based on the experiences and challenges faced by individuals with PWS and their primary caregivers. Therefore, to identify the most effective interventions, by generating more strategies and by using and modifying, where necessary, existing therapeutic methods.

### **1.10 Research approach**

This study will be achieved through qualitative research. According to Stiles (2003) qualitative methods are particularly well adapted for studying human experience and meaning. Qualitative data puts emphasis on people's lived experiences and is well suited for locating the meanings people place on events, processes, and structures of their lives, their perceptions, assumptions, prejudgments, presuppositions and for connecting these meanings to the social world around them (Miles & Huberman, 1994). For the purposes of this study, case studies with elements of ethnography,

auto-ethnography and participatory action research will be used. These will be discussed in detail in Chapter two.

### **1.11 Research theory**

Theory for this research will be discussed in detail in each chapter, when presenting the case studies. Applicable theories on developmental psychology and on therapeutic approaches will be discussed as relevant to each participant.

### **1.12 Ethical considerations**

Approval was obtained from the chief executive officer of the Dr. George Mukhari Hospital, and from the Head of the Clinical Psychology Unit where the research was conducted (see appendix II). Ethics clearance was obtained from the Ethics committee of the Department of Psychology at the University of South Africa, prior to conducting the research. Permission to conduct the study with registered members of the Prader-Willi Syndrome of South Africa Association was obtained from the Association and from its members (see Appendix I).

Prior to conducting the research, participants were given verbal information about the purpose and nature of the research project. This was achieved by giving a brief presentation at the Association's yearly information session. Participants were informed that all interviews would be audio-taped, that they were free to participate or decline to participate, and that they needed to give written consent (HPCSA, 2002) (see appendix III). Participants were assured that their information would be treated confidentially (Bless & Higson-Smith, 2000).

### **1.13 Presentation**

- *Chapter one* describes Prader-Willi Syndrome, physical symptoms, and cognitive impairment and behavioural problems, including food-related behavioural problems; treatment options are also discussed.
- *Chapter two* expands on the Prader-Willi Syndrome literature.
- *Chapter three* discusses the research methodology.
- *Chapters four to eight*, present the case studies.
- *Chapter nine* presents the conclusions, evaluations and recommendations.

#### **1.14 CONCLUSION**

This chapter discussed the physiological make up of Prader-Willi Syndrome, behavioural and psychiatric problems associated with this genetic disorder, as well as treatment. Based on the discussion, the significant impact the disorder has on the individual and the primary caregiver was highlighted and therefore it is essential to develop effective and adequate therapeutic methods. The research methodology and ethical considerations were also highlighted in the chapter.

## **CHAPTER 2**

### **LITERATURE REVIEW ON PRADER-WILLI SYNDROME**

#### **2.1 Introduction**

In Chapter one, Prader-Willi Syndrome (PWS) was introduced, and in this chapter further information on PWS is provided. In so doing, there is a focus on advances in the clinical management of the condition, research on medical illnesses associated with PWS is explored, and information on parental experiences is presented. It is also argued that, as with most conditions, a diagnosis is only the beginning of treatment: on-going management of the individuals and their families is essential.

#### **2.2 Early development in PWS**

##### **2.2.1 Infancy**

PWS is characterized by hypotonia especially in infancy, and as a result its presentation can be mistaken for other disorders like Werdnig-Hoffman (a rare form of spinal muscular atrophy presenting in infants), or Trisomy 18 syndrome (a chromosomal disorder caused by an extra chromosome 18), or a brain anomaly (Butler, Hanchette & Thompson, 2006). Subsequently it is likely to be misdiagnosed, and this may delay proper management. In most cases hypotonia is present from birth to 9 months (Tauber, et al., 2017). Because of poor suck reflexes, special feeding techniques are usually required to provide adequate nutrition, and, as a result a prolonged hospital stay is necessary (Cassidy, 1988).

Temperature instability (hyperthermia) is another characteristic which may be present during early infancy with high or low body temperatures. The bouts of hyperthermia or fever may trigger medical investigations to find the cause of possible infection, which is not often found (Butler et al., 2006).

In addition to the features mentioned in Chapter one, Butler et al. (2006) further described the mild dysmorphic features recognized during infancy which include a narrow forehead; a long, narrow appearing head; a small, upturned nose; a thin upper lip; sticky saliva; and down-turned corners of the mouth. PWS individuals may have diminished facial mimic activity due to hypotonia, as they grow older. They may also present with hypo-pigmentation, and this suggests that compared to other family

members they may have a fair skin and light hair colour during infancy. They furthermore lack eye co-ordination (strabismus), which means that the child's eyes may not move together and cross or wander to the side. This is usually correctable through patching or surgery at an early age.

### **2.2.2 Caring for an infant diagnosed with PWS**

Once the diagnosis of PWS has been confirmed, it is vital to ensure that the infant gets adequate nutrition, because infants with PWS do not spontaneously demand feedings, as they rarely wake up to feed. Therefore, a regular feeding schedule should be established and the infant's diet must be adjusted, as needed, to maintain appropriate weight gain (Cassidy & Driscoll, 2009).

Because PWS infants have poor feeding because of weak suck reflexes and low muscle tone, they need assisted feeding for the first four to six months. It is often necessary to encourage the infant to suck by stroking the cheek or gently squeezing the cheeks together. In the past, some parents have fed new-borns with a dropper or spoon to ensure adequate calorie intake, while many have chosen to introduce cup feeding early in order to circumvent the inadequate suck reflex (Cassidy, 1988). In recent years affected children could benefit from using a nursing system with one-way valves and manual sucking assistance, which was originally designed for infants with a cleft palate. Making use of this will reduce reliance on feeding tubes (McCandless & The Committee on Genetics, 2011).

Nasogastric tubes can also be used when needed, as they are generally well tolerated and are only required for three to six months. The use of a gastrostomy feeding tube can be considered, but can usually be avoided. Such a feeding tube is inserted through the abdominal wall and into the stomach, and allows the child to be fed directly into their stomach bypassing the mouth and throat. However, after considering the risks and benefits, it should be removed when it is no longer needed (McCandless & The Committee on Genetics, 2011).

Cassidy and Driscoll (2009) stated that for further management it is important that calorie intake be assessed and that weight and head circumference are plotted on growth charts monthly for the first six months. Early infant stimulation programmes



have to be implemented to optimize strength and milestone achievement. Encouragement of physical activity is important for strength and alertness at all ages.

Tauber et al. (2017) conducted a study where they administered intranasal oxytocin (OXT), a neuropeptide that plays an important role in modulating social interactions and mother-infant bonding. The study proved that a short course of repeated intranasal OXT administration is well tolerated and improves oral feeding and social skills in infants with PWS. After OXT treatment, infants were more alert, less fatigued, more expressive, and had less social withdrawal. They initiated mutual activities and were more engaged in relationships through gaze, behaviour, and vocalization. These modifications helped the parents to be more sensitive. The mother-infant dyad was less restricted and there was a better reciprocal exchange, thus engaging the dyad in a positive transactional spiral as well as optimizing feeding.

### **2.3 Early childhood**

By eighteen months to two years of age the feeding behaviour of children with PWS changes radically and an insatiable appetite may develop which could cause major somatic and psychological changes in early childhood (Butler et al., 2006). As the child with PWS grows, there is a progression of behavioural issues.

Butler et al. (2006) established that lying, stealing, and aggressive behaviour are common during the childhood years, and continue into adolescence and adulthood. Children with PWS do not easily agree to anything, are resistant to new ideas and experiences, and are more dependent than typical developing children. In addition, they are less physically active than their peers. Another identified aspect is lack of emotional control in both children and adults with PWS. This lack of emotion could be attributed to the dysfunctional hypothalamus, which is the control centre of hormonal functions and emotional stability.

### **2.4 Adolescence and adulthood**

Normal puberty is absent or delayed in both genders with PWS, while adolescents and young adults appear young for their chronological age, and all this result in reproductive problems. In the United States of America, only two women who were

presumed to have PWS in the 1970s became pregnant and one woman was pregnant on two occasions. However, the diagnosis of PWS was not confirmed with genetic testing in either of them (Butler et al., 2006).

Most recently, at least three women with confirmed PWS have been reported pregnant. In these cases, one woman was 33 years old in Sweden and gave birth to a healthy girl by caesarean section delivery after an estimated 41 weeks gestation. The other adult female gave birth to an infant with Angelman syndrome (Akefeldt, Tomhage & Gillberg, 1999; Butler et al., 2006). Angelman syndrome is a genetic disorder which primarily affects the nervous system. Features include: delayed development, intellectual disability, severe speech impairment, problems with movement and balance, recurrent seizures, and small head size (Adams, 2008).

Even though they may have hypogonadism, it is important to recognize that teenagers with PWS deal with the same hormonal issues that all adolescents encounter and with their typically developing peers, many of their behavioural issues seem to stabilize, although they do not disappear entirely, as they reach adulthood (Honey, 2010; McCandless & The Committee on Genetics, 2011). Typical behavioural problems include rigidity of personality, perseveration in conversation, tantrums, and non-compliance, and occasionally worsen during adulthood (Butler et al., 2006).

Whitman and Jackson (2006) reported that a survey of parents from the United States of America, Australia and New Zealand found that conflict between parents contributed to severe behavioural problems. Any disagreement and lack of a common goal will confuse the person with PWS especially when one parent or family member is saying the person can do one thing, while another is saying the opposite. The individual with PWS needs consistency and also needs to learn certain behaviours from other family members. This aspect will be elaborated on, later in this chapter.

Young adults with PWS seem to be prone to a variety of compulsive behaviours, and some of them develop obsessive-compulsive disorder. Some behaviours that are classified as obsessions are: distress when changing routine; repetitive questioning;

hoarding; need to tell or ask; excessive showering, toileting, or grooming; repeated organization, writing, or collecting; and the need to finish one thing before moving to the other. A significant majority of young adults with PWS develop depression, anxiety, and sometimes psychosis. Parents should be counselled to identify early indicators of these processes to facilitate appropriate medical intervention (Cassidy & Driscoll 2009; Honey, 2010; McCandless & The Committee on Genetics, 2011).

Rocha and Paiva (2014) reported results of a comparative study investigating the prevalence and severity of obsessive compulsive symptoms and Prader-Willi-like phenotypes (the term PW-like is used to refer to a patient with clinical features that are very similar to PW, but without the confirmation of a classical genetic subtype that can cause the syndrome). They showed that PWS patients suffered higher incidents of obsessive-compulsive symptoms and more severe symptoms compared to their PW-like counterparts.

Butler et al. (2006) discovered that when an adult with PWS is surrounded by caregivers who have been trained in the management of PWS, behaviours can be managed. When the environment is one in which caregivers are unaware of the specific needs of adults with PWS, behavioural deterioration can be expected.

Psychologists can help families during this stage by explaining the exact nature and implications of PWS to the affected individual, extended family, friends, and baby sitters. Psychotherapeutic group sessions and couples therapy are indicated in some cases and also small group sessions for siblings from several families in order to assist the siblings understand the genetic implications of the disorder and to overcome embarrassment, express their anger in constructive ways, and promote cohesion (Sulzbacher, 1988).

## **2.5 Diagnosis of PWS in adults**

Scheemeyer (2013) reported that each year new diagnoses are made in patients who are in their 20s and 30s. Many such patients have been misdiagnosed, and have been given alternative diagnoses like intellectual disability, Asperger's syndrome, autism spectrum disorder or even other chromosomal abnormalities such as a PWS-like subtype of the Fragile X syndrome.

The General Practitioner or allied health professional can play a major role in identifying adults with PWS, by re-assessment of an existing clinical diagnosis. For example, if an adult patient is short, obese, has intellectual disability or learning difficulties, and also has hypogonadism, small hands and feet, and insatiable appetite, and thick, viscous saliva a re-investigation for PWS is warranted. In addition, a history of general feeding problems in early infancy that may have required tube feeding can often be considered confirmation of a clinical diagnosis of PWS. Speech and articulation problems, as well as skin picking, are other highly characteristic features of PWS that need to be investigated at this age (Scheemeyer, 2013).

## **2.6 Perseveration**

Whyte (1997) wrote about the continuous debate on whether the phenomenon of perseveration is a behavioural problem or a linguistic one. Some parents and teachers reported feeling personally attacked when they have answered the same question six or even sixteen times, and then the child asks it again. Whyte (1997) suggested the following interventions:

- 1) Diversion: where one can change the subject.
- 2) Reassurance: try confirming that the individual already knows the answer and s/he must tell you what it is. In this way, s/he can confirm that the answer is what s/he thought and this also indicates that further repetition is not necessary.
- 3) “I don’t know”: can also be an answer, but some children may have to be taught this concept. They may think that you are just not bothered or that if they ask again you will know next time.
- 4) Wait: if you cannot answer for some reason at that moment, you can mention a time by which you will answer.
- 5) Demonstrating knowledge: sometimes the repetition is just to let you know, or get you to confirm something s/he is not sure of.

If perseveration is regarded as a linguistic or higher language difficulty, the most effective way to bring about lasting change is to teach the correct, acceptable social exchange patterns so that a reference is available the next time they repeat it. It is not enough to explain to a PWS individual that it is upsetting to have the same

question repeated; if they are stuck on an idea, there is no way they can go on to another subject without help (Whyte, 1997).

## **2.7 Eating behaviour and obesity**

Butler et al. (2006) reported that during late adolescence some individuals begin stealing from food stores and rummage through discarded lunch bags or trash cans to find partially eaten leftover food or even inedible food items. Without intervention, adolescents may weigh 250 (113 kg) to 300 (136 kg) pounds by their late teens, which can lead to a shortened life due to the complications of obesity. Overeating can lead to immediately life-threatening events such as stomach rupturing. In the past many individuals with PWS died before the age of 30 years.

The primary health issues in individuals with PWS are exacerbated by obesity. When weight is kept under control, there are fewer serious health issues. When weight increases, disease associated with obesity may appear. Morbid obesity often causes obesity related hypoventilation, which can be a serious problem and demands attention (Butler et al., 2006).

Type 2 diabetes mellitus is diagnosed in 25-30% of PWS adults who become morbidly obese. Diabetes becomes difficult to control with medication if food restriction is inadequate. When food intake is reduced to the appropriate number of calories, diabetes usually comes under control quickly. If this cannot be achieved, complications of diabetes may occur within a few years after it is diagnosed. These may include retinopathy, neuropathy, kidney failure, and amputations (Butler et al., 2006).

### **2.7.1 Managing eating behaviour problems**

Research on eating behaviour in PWS has not resulted in any treatment that enables the individual to control their own eating behaviour. Obesity has been controlled using surgical intervention to date. At times medication is used, but these approaches do not solve the problem. The only identified effective way to control obesity is by restricting access to food. Parents could thus monitor food intake (Honey, 2010; Whittington & Holland, 2010). Hospitalization has been considered the only alternative with cases of severe obesity. A person may need to be admitted to

hospital to reduce weight under supervision (Waters, Jewson, Quinn & Sharma, 2007). Parents should be counselled that offering food as a reward or withholding food as punishment is almost always counterproductive and should be avoided (McCandless & The Committee on Genetics, 2011).

Nutritional counselling for long-term weight management should begin in early infancy in order to prevent inappropriate weight gain. Care should be taken to not over feed the infant with PWS. The lack of satiety should be acknowledged at school, work and at home and all caretakers and supervisors need to understand these factors. Consistent limit setting and close supervision are necessary at all ages (Cassidy & Driscoll, 2009). Designing diets high in bulk and low in calories has also been considered effective (Whittington & Holland, 2010).

## **2.8 Growth hormone therapy (GHT)**

Goranson (2011) reports changes on the use of growth hormone therapy. After years of debating about whether children with PWS have a growth hormone deficiency, doctors and researchers have found that the deficiency of growth hormone secretion probably originates in the hypothalamus. The hypothalamus has been identified as being responsible for different aspects of PWS, and plays a key role in growth and sexual development; it regulates appetite, metabolism, body temperature, and mood.

For the past 10 years the use of GHT has become part of the management of individuals with PWS (Goranson, 2011). A child with PWS can be assessed for GHT at any age. Clinical experience suggests that the treatment can be beneficial for an individual with PWS as early as two to three months of age (McCandless & The Committee on Genetics, 2011).

As stated in chapter one, GHT in children may assist with height, weight, body mass, strength, respiratory function, agility, and cognitive development. In addition, using a low dose of GHT in adults has led to positive results in bone strengthening and the promotion of leaner muscle mass and greater energy. Doctors, researchers and patients continue to report many dramatic changes and positive outcomes from the use of GHT in PWS individuals of all ages. There is no longer any doubt that GHT can improve the health and quality of life of individuals with PWS (Goranson, 2011).

Bakker et al. (2017) studied 522 prepubertal children with PWS treated with GHT for three consecutive years. After one year of treatment the height had improved and after three years their height had completely normalized.

According to Dykens, Roof and Hunt-Hawkins (2017) GHT is associated with improved cognition and adaptive behaviour that was sustained in children treated over time. One study compared untreated controls and adults who were on GHT for six months and found that adults on GHT improved in cognitive tests of mental speed, flexibility, reaction time, and motor performance. They also showed improved aerobic conditioning, muscle mass, strength, attention span, energy, and well-being. The study also found that continuously treated versus untreated children maintained their advantages over time in verbal and full-scale IQ scores, and in their adaptive communication and daily living skills. Although GHT is associated with cognitive benefits, a longer treatment period does not necessarily yield increasingly higher IQ scores.

One of the challenges of GHT is that the caregivers must learn to give the injections at home. The shots are usually given at night, six times a week, by the caregiver or the individual himself/herself. Night-time is recommended, because the largest natural spurt of growth hormone release occurs in the first few hours of sleep (Goranson, 2011).

As with any medication, GHT may cause undesirable side effects in some cases. The most common side effects are minor, such as changes in the skin at the injection site, for example, occasional bruising, slight bleeding, tiny bumps on the skin, or an indentation. Goranson (2011) also reported individual experiences that required attention like headaches, swelling of feet and legs, increased insulin levels, decreased thyroid hormone levels, respiratory dysfunction, progression of scoliosis, elongation of the lower face, and extreme overgrowth.

## **2.9 Sleep problems**

Sleep problems are commonly observed and are reported to be one of the characteristics in individuals with PWS. These include excessive daytime sleepiness, day naps after five years of age, sleep apnea (a sleep disorder characterized by

pauses in breathing or instances of shallow or infrequent breathing during sleep), restless sleep, and excessive night-time sleep. It has been reported that 72% of individuals with PWS suffered from day time sleepiness, 66% slept more than 8 hours per day, and 44% suffered from loud snoring during sleep (Cassidy & Driscoll, 2009; Honey, 2010).

## **2.10 Speech and language disorders**

The speech and language skills of individuals with PWS are reported to be below expectation, based on intellectual levels. The difficulties include poor speech-sound development which includes errors due to poor motor abilities associated with the production of speech-sounds and errors in applying linguistic rules to combine sounds to form words; reduced oral motor skills; and language deficits. Language problems include deficits in vocabulary, grammar, morphology, narrative abilities, and pragmatics (Lewis, 2006).

Several factors may account for the poor speech-sound development of individuals with PWS including oral structure abnormalities, abnormal saliva, hypotonia, poor phonological skills, and cognitive deficits. It is more likely that poor oral motor skills, especially reduced tongue elevation for speech and slower alternating movements of the articulators, account for poor speech-sound skills in PWS individuals. Speech-sound errors in individuals with PWS include sound distortions and omissions, vowel errors, simplification of constant blends, and difficulty in sequencing syllables (Lewis, 2006).

Lewis (2006) found that some individuals demonstrate atypical patterns such as a phonological disorder or apraxia of speech. Apraxia of speech is a severe speech-sound disorder that includes impairments in syllable sequencing, prosody, and speech-sound characteristics. The voice of individuals with PWS may differ in pitch, quality, intensity, and resonance, compared to those of their peers. Voice characteristics reported for individuals with PWS include a high-pitch voice, harsh/hoarse voice quality, inadequate vocal intensity, and hyper nasality.

According to Lewis (2006) a few studies have examined narrative skills of individuals with PWS. Both children and adults with PWS were observed to have had difficulty



with story retelling tasks. While narrative skills appear to develop into adulthood, the narrative abilities of the individual with PWS lag behind other language skills. Poor narrative skills may contribute to deficient conversational skills and thus impact on social skills, interpersonal relationships and job-related communication skills. Pragmatic deficits, including problems with maintaining a topic, judging appropriate proximity to the conversational partner, and turn taking, have also been observed. The pragmatic deficits may impede progress in therapy (Lewis, 2006).

Surprisingly, despite oral language deficits, individuals with PWS show relative strengths in written language skills. The speech language pathologist can become involved with the child with PWS soon after birth. Children begin acquiring word understanding essentially from birth. Among typically developing children, expressive language follows soon after birth with cooing at three months, babbling at six months, and consonants in the form of “dada” and “mama” at around eight months. By ten months of age the consonants are used discriminatively, and there is evidence that the child understands the meaning of the word “no”. By twelve months of age, most children have acquired two words, in addition to the consonants (Lewis, 2006).

Children with PWS are, however, eighteen months of age before they begin to acquire verbal vocabulary. A substantial number of affected children acquire speech much later some as late as six years of age. Pragmatic difficulties may be noted due to poor social skills and the emergence of behavioural disturbances. If the child’s speech is highly unintelligible, ACC (augmentative and alternative communication) may be considered. The ACC system allows an individual to build vocabulary and pragmatic language skills while oral skills are developing. ACC includes sign language or communication boards. It is usually transitional until oral speech abilities improve, and alleviates some of the frustrations that the child and caregiver may experience. At school age (six to twelve), children with PWS are usually enrolled in speech and language therapy through the school they attend. Emphasis should be placed on functional language skills and life-skills training (Lewis, 2006).

## **2.11 Other medical illnesses**

During adolescence and adulthood, PWS afflicted individuals usually have to cope with the various health problems described below (Butler et al., 2006; Honey, 2010).

### **2.11.1 Patella-femoral syndrome**

According to Goelz (2006) patella-femoral syndrome is a common cause of knee pain that is grossly under-reported in all teens. Even when musculoskeletal development and lean body mass are within normal limits, muscle and soft tissue imbalances at the hip and knee are a frequent cause of patella-femoral knee pain. The hypotonia and typical body shape render the teen and young adult with PWS even more susceptible to this very common cause of knee pain. Patella-femoral syndrome results when the patella or kneecap does not accurately track through the patellar groove during flexion and extension movements. The faulty tracking occurs as a result of a variety of musculoskeletal factors. Patella-femoral syndrome occurs when bending or straightening the knee. Those troubled by the syndrome will experience pain when ascending and descending stairs, and will frequently describe knee pain upon rising after extended periods of sitting (Goelz, 2006).

### **2.11.2 Scoliosis**

Individuals with PWS are more at risk of neuromuscular scoliosis, presumably as a result of low muscle tone. Approximately 62-68% of the PWS population have scoliosis, with a structural change of at least 10 degrees. The higher prevalence of neuromuscular scoliosis among children and teenagers with PWS necessitates careful monitoring by radiographic studies especially during periods of rapid growth. The treatment of neuromuscular scoliosis ranges from careful monitoring, through to bracing, and surgical stabilization (Goelz, 2006).

### **2.11.3 Epilepsy**

The frequency of epilepsy in PWS patients is higher than in the general population, ranges from 4-26%. The types of seizures include generalized tonic clonic; complex partial; atypical absence; staring spells; and myoclonic, tonic and hemiclonic seizures; but the most frequent type is focal epilepsy. However, generalized and focal seizures are the most frequently reported findings. Epilepsy in PWS is usually

responsive to antiepileptic monotherapy with rapid seizure control and a good outcome (Verrotti, Soldani, d'Alonzo, & Gross, 2014).

### **2.12 Self-injurious behaviour**

Skin picking, which has been highlighted earlier, and other forms of self-abusive behaviour are found in most individuals with PWS. Serious health problems deriving from persistent self-injury may occur, and include eye poking, subdural haemorrhage from forceful head banging, infections from self-inflicted skin picking, and anorectal disease from rectal picking and digging. Other forms of self-abusive behaviour have been reported and include trichotillomania, pushing pins and tacks into the skin, and pulling out nails (Verrotti et al., 2014).

### **2.13 Life expectancy**

The natural life span of individuals with PWS is currently undefined. Some individuals live into their seventh decade of life. An individual who died at the age of 71 years was described in 1994, and a second individual aged 68 years was described in 2000. The PWSA (UK) is also aware of a woman with PWS who recently died at the age of 74. However, experience suggests that survival past the fifth or sixth decade is unusual. For instance, in a survey of 232 adults with PWS, the oldest was 62 (Eiholzer & Lee, 2006).

A significant number of people with PWS continue to die in their 20's, 30's and 40's. A review of causes of death in individuals with PWS in 1996 indicated that obesity-hypoventilation syndrome (a breathing disorder in which poor breathing results in too much carbon dioxide and too little oxygen in the blood) caused most deaths. If obesity is controlled and overeating avoided, life expectancy should be prolonged (Butler et al., 2006).

### **2.14 Education and social issues**

Taylor (1988) noted that the IQ of individuals with PWS often decreases during the early years, particularly between the ages of six and ten years. A child's IQ decreases over time, and this does not imply that the child is losing cognitive skills as he/she gets older. Most of these children learn a significant amount of information and become more competent in terms of daily living skills. The decrease in IQ

suggests that as the child with cognitive problems gets older the discrepancy between his/her performance and the average performance gets larger. For one thing, as the child gets older, his/her IQ is based on more and varied tasks. This could explain the greater decline in IQ between the ages of six and ten years.

According to Goff (2006) many children with PWS begin school in mainstream settings. About 5% attend regular school until secondary level but the intellectual impairment and potential behavioural problems present in most children with PWS by this age, require special education and support services. The transition to a special school needs to be carefully planned, teachers need to be prepared to work with the student with PWS, and the student may need reassurance that the school change is not the result of some kind of failure but is rather an opportunity to learn better and to make new friends (Goff, 2006).

A psychologist's role is important during the schooling years. Establishing a general intelligence level is important as early as during the pre-school years, and appropriate educational placement requires psychological evaluation and recommendations. In most cases, once the transition to elementary school is completed, the need for psychological services diminishes, until the child is about ten years old. When the child is in junior high, educators frequently seek psychological advice (Sulzbacher, 1988).

Chedd, Levine, and Wharton (2006) advised that educators need to be aware of several medical and psychological features associated with PWS particularly an altered level of arousal, the pronounced appetite disturbance, and diminished muscle tone and motor planning skills. These features impact on both classroom performance and perceptions of children by teachers and classmates.

School aged children with PWS usually do well in regular classrooms when provided with extra services. Another approach involves regular morning kindergarten classes and a special language-based classroom in the afternoon that allows for individualized teaching. This format may be quite successful, and can be continued throughout the elementary grades (Chedd et al., 2006).

In addition, the classrooms need to be modified by including chairs that provide back support and which are of appropriate height. Desk and table tops should be at below height bearing in mind that PWS children are very short, ensuring that the work surface is an easier and less fatiguing work space (Minor & Carr, 1988).

Goff (2006) found that the transition from elementary school to middle school, and again to high school, poses dramatic changes. The safety and security of being in a single classroom in a familiar building with the same teacher for several years is suddenly taken away, and classmates who have been together for many years are separated. For some students, this is an exciting opportunity to be independent and to grow up. However, for individuals with PWS, it may be the beginning of loneliness and rejection.

Adolescents with PWS desire the same things as other teenagers: friends and close relationships. They can, however, carry this desire to extreme lengths in their search for a boyfriend or girlfriend including obsessing over a particular individual. They need education and guidance in this area. Rules and guidelines may be needed that outline where and when a public display of affection can take place. They may also need to learn what should be said or done to make sure that consent is obtained (Goff, 2006).

Adolescence is traditionally a challenging time, with children having to cope with increased pressures coming from all directions. For those with PWS, the growing awareness of the differences between them and their non-PWS peers, occurs at the same time that being “just like” one’s friends is so important. Individuals with PWS who observe changes in their peers (but none in themselves) are likely to demonstrate increased stress which may create anger, resistance, and food-seeking behaviours that interfere with learning and adaptation. The school can benefit from regularly scheduled parent conferences and outside consultation from a professional familiar with PWS (Chedd et al., 2006).

Due to behavioural challenges, and regardless of their cognitive functioning, most PWS children are not ready for the full autonomy their same age peers are achieving, and most need continued supervision and protection. Both teachers and

parents need to provide extra security in the school and home while seeking ways that adolescents with PWS can increase independence, participate in enjoyable activities, and improve their quality of life. School programmes should capitalize on the individual strengths and teach coping skills for dealing with challenges (Chedd et al., 2006).

While challenges exist, there are positive aspects during adolescence. Many teenagers develop effective verbal skills and become active participants and contributors to school activities. This is a time during which vocational planning and work experiences should begin to be pursued. While it is generally difficult for individuals with PWS to foster friendships independently, satisfying relationships are possible especially if parents, teachers, and other supportive adults in the community are involved (Chedd et al., 2006).

For most individuals with PWS, formal education ends between the ages of 18 and 21, but if vocational training has been successfully introduced before that time, a smooth transition to the world can occur. Unfortunately, many individuals do not have this opportunity, and there is a gap between completion of school and entrance into a job-training programme. This loss of daytime structured activities usually results in behavioural deterioration and increased health problems (Butler et al., 2006).

### **2.15 Vocational training**

When the child with PWS reaches high school age, prevocational assessment can provide the foundation for vocational training for employment once the formal education process is completed. An assessment by an occupational therapist is essential. Areas to consider in assessment of prevocational skills include posture and movement, hand/arm use, and work habits (Minor & Carr, 1988).

### **2.16 The continuum of work placement options**

Drago (2006) stated that sheltered workshops are the oldest and most common type of job placement for workers with developmental disabilities. These settings offer job skills training in a non-integrated setting. This means that the entire work or training force comprises disabled individuals. Typically, sheltered workshop tasks include

packaging and simple product assembly. Individuals in sheltered workshops do not receive an hourly rate, but are paid for each piece of completed work at a rate comparable to that paid to a non-disabled person.

Other opportunities include work enclaves which consist of small, non-integrated groups of disabled individuals who go into the community to perform service type work. Typical work enclave jobs are lawn maintenance, and janitorial and restaurant work. Each enclave has at least one non-disabled supervisor. Individuals in work enclaves are paid an hourly rate (Drago, 2006).

Drago (2006) added that individuals with PWS can get involved in supported employment, which is a community based job placement in an integrated work force. The other workers performing comparable jobs are non-disabled. Individuals placed in supported employment must receive a wage that is equal to or above the minimum wage and comparable to that of non-disabled individuals performing the same job at the same location. Supported employment is supervised by a job coach, who provides onsite job training and acts as a liaison with the employer. Ultimately the goal is for the job coach to become less necessary, until, ideally, the job coach is no longer needed. When this occurs, the individual is said to be competitively placed. Individuals can progress from a sheltered workshop to an enclave, to supported employment, and finally to competitive employment (Drago, 2006).

### **2.16.1 Challenges to successful vocational placement**

Drago (2006) stated that vocational placements are still difficult to achieve for individuals with PWS. There are several reasons for this. First, vocational providers have not had to live with the individual with PWS; many are slow to understand the seriousness of their appetite and the emotional volatility associated with the condition. When an individual with PWS disappears from the work site to obtain food or becomes argumentative with the employer, employers will most likely perceive these behaviours as discipline problems rather than as a natural manifestation of PWS which requires workplace adaptation. Another issue encountered was getting individuals to work on time; getting up and out of the house is often difficult for individuals with PWS. Other problems have occurred around returning from

bathroom breaks on time, and disengaging from one work task and beginning another (Drago, 2006).

Kazemi and Hodapp (2006) in a telephonic survey with parents in the United States, found that when it came to vocational issues, the major concern was the lack of knowledge of PWS. The debilitating nature of PWS is often difficult to grasp until time is spent working or living with an affected individual (Drago, 2006). Staff working with PWS individuals get frustrated and have a low tolerance for various associated behaviours. Behaviours characteristic of the syndrome frequently lead to job terminations (Drago, 2006; Kazemi & Hodapp, 2006). Based on this challenge, parents need to educate adult service staff about PWS and its effects. What was also found to be useful was having the individual's special education teacher assist with adult services, given they knew the individual well (Kazemi & Hodapp, 2006).

Many individuals with PWS select work in a lawn crew, as one of their goals. Lawn work is hard physical labour. Many currently employed adults with PWS reached adulthood before growth hormone replacement therapy was available, and as a result, they may lack the physical stamina required for lawn work. In addition, the summer sun may be medically contraindicated for adults with hypopigmentation (Drago, 2006).

The supported employment which has been identified as another vocational option could be a problem because of the unrestricted nature of the environment along with the lack of supervision. For many in these settings, maintaining dietary restrictions must rely primarily on self-control. To ensure employment success, employers must be aware of and address these issues prior to placement, must provide constant monitoring of these issues after placement, and, finally, must be prepared to deal with the challenges if and when they occur (Drago, 2006).

Another issue is that many individuals with PWS may not quickly or easily adapt to a supported employment setting. As a result, rehabilitation counsellors and job coaches might be dismayed by the increased tantrums and disruptive behaviour shown by young adults with PWS during the first few days or weeks on the job. The first reaction of these professionals may be to terminate employment, and to assume



that the individual cannot possibly handle the demands of a particular work setting. Given time, the individual may in fact get used to job demands and routines. Parents may need to work hard to help the manager, rehabilitation counsellor and job coach to expect a slower, more gradual transition into an appropriate work placement (Kazemi & Hodapp, 2006).

## **2.17 Residential care for adults with PWS**

Provision of residential services to adolescents and adults with PWS was not seriously addressed prior to the late 1970's, primarily because until then most individuals were not expected to live beyond their adolescent years. As a result of earlier diagnosis and appropriate nutritional and weight management, individuals with PWS now live well into adulthood (Ziccardi, 2006).

Many parents noted there were not enough Prader-Willi group homes. At the most basic level, placements are difficult to locate and secure, and subsequently many parents were forced to choose between several less than perfect options and for many families exploring residential placement for their family member is an overwhelming and emotionally charged task (Kazemi & Hodapp, 2006; Ziccardi, 2006). Many families face long waiting lists and lack governmental funding for developing appropriate, alternative living opportunities. Families are often forced to wait years for any residential facility that is even remotely willing to assume the challenges of serving someone with PWS (Ziccardi, 2006).

When they finally secure a placement, parents have been concerned about the frequent staff turnover in group homes where their children reside. Because of such frequent changes, it is often difficult for parents to develop relationships with staff members or for staff to get to know the individual with PWS. Several parents recommended that in order to get a balanced view parents should speak to both the staff and the individual about the progress. Parents might also speak to the group home counsellor, so that they can get an overview of group home activities (Kazemi & Hodapp, 2006).

The family's full participation and disclosure of their family member's strengths and needs is the first step in securing a successful residential placement. A residential

provider has a significantly improved chance of meeting the individual's needs, if those needs are discussed honestly and openly. Many willingly provide behavioural accommodation and environmental modifications to facilitate a lasting placement that includes a clinically sound approach to treatment, an acceptable quality of life, and positive outcomes. The prospective provider and family will be most successful for the individual with PWS, if a spirit of openness and cooperative, respectful communications are established early in the relationship (Ziccardi, 2006).

Several parents have noted problems their child had when outside in the community. The most persistent issue has been manipulation and the ways that peers take advantage of young adults with PWS. In reacting to this issue, parents found that the solution requires a delicate balance between the parents or group home staff's desire for protection, and the young adult's need for independence (Kazemi & Hodapp, 2006). Families and providers also need to openly discuss issues regarding their views and normalizing the environment for the PWS individual and to what depth community integration is expected (Ziccardi, 2006).

Environmental factors are also important for providing a safe and successful residential programme for people with PWS. For most, securing food is primary, as to individuals with PWS the knowledge that food is inaccessible is a comfort and reduces much anxiety (Ziccardi, 2006). Many residential programmes choose to utilize a totally locked kitchen; others elect to lock refrigerators and cupboards; either method is appropriate. Most importantly, consistency in properly securing the locks must be applied by everyone at all times. A menu board posted daily may help alleviate some of the ever-present questions and concerns regarding food. To accommodate the short stature of many individuals with PWS especially those who have not benefited from GHT, providers may want to lower shelves and closet rods when building or modifying a home (Ziccardi, 2006).

Individuals with PWS have unique space needs. They have a tendency to both collect and hoard, and have an extraordinary sense of protectiveness for these collections particularly in relation to others. This suggests that the resident must have ample space (Ziccardi, 2006). Individual bedrooms which are locked and which are accessible only to the occupant and support staff, are ideal for reducing both real

and imagined incidents of stealing and property destruction. When single bedrooms are impossible, strict guidelines need to be established and enforced with regard to individual space, property, and privacy. Integrating these expectations into the house rules may help prevent disagreements later on (Ziccardi, 2006).

A successful residential programme requires well trained, supported, and empowered staff. These attributes are even more critical for supporting people with PWS. Furthermore, staff working with individuals with PWS must be mature, must display consistency and not need the last word in a debate. Finally, staff must demonstrate forgiveness and know that regardless of what happened today, tomorrow is a new day. The content of the programme is as critical as staff training is. People with PWS require consistency and structure in all areas, including programming. Generally, a basic set of house rules, applied equally to all, will establish baseline standards for acceptable behaviour. Behaviour support plans must be individualized, and must address the unique needs, strengths, and desires of each person (Ziccardi, 2006).

The provider and parents must work to develop relationships with physicians willing to provide primary care as well as specialists for psychiatric oversight, podiatric care, dental treatment, and a host of other speciality areas. A nurse employed by the residential provider can have a positive impact on the relationship between physicians and individuals requiring medical care. The nurse also plays a critical role in educating others including neighbours, teachers and vocational support staff (Ziccardi, 2006). An approachable, informed dietician is essential to create menus, discuss food preferences, and to determine what food items are permissible for special occasions. It is beneficial that staff receive training related to the food aspects of the syndrome directly from the dietician. An exercise physiologist also has a major role in a residential programme. The exercise physiologist must assess each individual's overall status, and once the assessment is completed an appropriate exercise schedule can be created and individualized (Ziccardi, 2006).

## **2.18 Parental experiences and concerns from birth to adulthood**

### **2.18.1 Birth**

Caldwell and Taylor (1988) gathered information from 12 families and found that at the time of birth a diagnosis of PWS was extremely rare. Until infants began to demonstrate the phenotypic characteristics some were misdiagnosed or not diagnosed at all. The children were given a poor prognosis with high probability of having significant cognitive limitations. Parents gave different views on how the birth of the child impacted on their marriage. Some parents indicated it had no impact, others mentioned that it strengthened their relationship as a result of mutual support, while others suggested it caused marital problems. The impact on the sibling was documented as having no impact by some, while others reported the children were drawn closer together or felt that the children felt frustration and embarrassment. At this stage parents needed information on the diagnosis, how to manage the child's unique problems, and on emotional support needed to help them cope with the pressures.

Whitman (2006) found that parents of children with PWS reported higher levels of parental and family problems, suffered greater pessimism, and yet got less support from professionals when compared with parents of similarly aged children with cognitive deficits from other causes. Whitman (2006) further highlighted that stressors of everyday life can greatly affect both general family functioning and specifically the ability of the family to adequately provide for their child with PWS. Having a child with PWS also places further stress on marriages. Harried schedules, work overload, financial difficulties and other external precipitants of stress may become overwhelming.

### **2.18.2 Weight gain**

As the child grew some parents were concerned about weight gain from six months, and two to three years old while others only became concerned at seven years. When faced with this challenge, some parents indicated they felt sorrow and others felt guilty and helpless regarding their child's weight gain. During this period parents still wanted a correct diagnosis and had needs for programmes or techniques to promote weight management. They also needed support from other parents (Caldwell & Taylor, 1988).

### **2.18.3 Adolescence and post-adolescence**

Parents were uncertain of the age at which their child entered adolescence. A relatively small percentage indicated that their children displayed typical physical signs of adolescence. Others reported that their child exhibited stubbornness, temper tantrums and other behaviours traditionally associated with PWS during the adolescence stage. Most parents reported that the diagnosis of PWS was made during this period. Also during this time most parents stated that their children were receiving some type of special education for behaviour disordered, learning disability or mildly retarded students (Caldwell & Taylor, 1988).

Interestingly, most parents noted that during this time period there was marital discord primarily related to depression, tension, and the uncertainty about the future of their child. There also seemed to be a significant amount of sibling problems. These problems ranged from jealousy to extreme protection. The resources that parents needed were financial information such as social security insurance, information regarding future living arrangements and information on weight and behaviour control (Caldwell & Taylor, 1988).

Many families could maintain a stable marital relationship, but fundamental differences in parenting styles were a constant source of conflict. Since children and adults with PWS have an extraordinary need for sameness and consistency, parenting style differences must be identified as early as possible and need to be sensitively addressed to prevent later behaviour problems (Whitman, 2006).

When parents are obliged to constantly focus on the needs of their affected child the risk for siblings having adjustment difficulties, although not inevitable, is clearly increased, and they often have ambivalent feelings towards their sibling with PWS. They may be embarrassed by the behavioural problems associated with food especially when they occur in public. Siblings often express reluctance to invite their friends to their home, while at the same time indicate a lot of guilt associated with this reluctance. Jealousy about the attention given to the sibling with PWS is often noted. Siblings are often pressured into parenting roles, or are given additional responsibilities (Whitman, 2006).

In addition, Thomson, Glasson, Roberts and Bittles (2017) found that carers of individuals with PWS reported high stress associated with the initial diagnosis. Their stressors also included lack of quality information about the disorder, time constraints and physical and emotional tiredness. Parents were also found to have adopted a variety of coping strategies such as learning about the disorder, accepting the situation, and seeking social support. Parents felt that accurate and timely information during the time of the diagnosis helped.

## **2.19 Behaviour management**

Sulzbacher (1988) emphasized that psychologists are faced with important issues when managing individuals with PWS. These include school placement, management of food-related behaviours, and problems like stubbornness and temper tantrums. The need for psychological advice seems to occur at predictable intervals as PWS individuals grow up. A request for a clinical re-evaluation or referral to a psychologist is generally precipitated by a behavioural crisis at home or at school.

### **2.19.1 Managing food-related behaviours**

Whitman and Jackson (2006) found that effective management requires the unyielding presence of four elements: (1) a physical environment structured so that food access is completely eliminated; (2) an appropriate dietary and exercise plan; (3) a procedure for ensuring that the affected person is always informed about the time and menu for the next meal or snack; and (4) elimination of all other avenues for obtaining food. Parents and caregivers reported that, over time, when the person with PWS realizes that excess food is no longer available many seem to experience a marked reduction in anxiety and report feeling safer, knowing the limits are effective and inviolable. While a family or care-giving setting may be able to control access to food, other environments where the individual with PWS may spend time are difficult to manage including schools, the work environment, recreational programmes, churches, and shopping malls. For this reason, there is a need for increased supervision to prevent food access (Whitman & Jackson, 2006).

### **2.19.2 General strategies and approaches to intervention**

According to Whitman and Jackson (2006) successful behaviour management requires the following:

- **A structured environment**

Much of our behaviour is influenced and controlled by the environment, and most individuals with genetic disorders have altered perceptions of the environment.

Those with PWS are extremely sensitive and reactive to the environment. In addition to heightened environmental sensitivity, persons with PWS tend to over-react to and overemphasize the negative aspects, while under-reacting to positive aspects of the environment. Therefore, the role of the surrounding environment in supporting or impeding positive behaviour management and positive adjustment for those with PWS, cannot be over emphasized (Whitman & Jackson, 2006).

A structured environment is more effective. The structure should include 1) family and house rules regarding what is expected, acceptable and unacceptable behaviour, together with specified consequences; and 2) schedules that are invariant in terms of time and task routines. Consistency between parents or caregivers is critical since inconsistency invites frustration and acting out. In addition, the structure must anticipate and provide carefully designed opportunities for the individual to make choices (Whitman & Jackson, 2006).

- **Strategies for changing behaviour**

Whitman and Jackson (2006) stated that the success of behavioural interventions hinges on the ability of caregivers to provide the necessary intervention with consistency and integrity. The most successful behavioural approach includes a general strategy of emphasizing positive social interactions and eliminating coercion. Examples of coercion include: questioning, arguing, threatening, using verbal or physical force, criticism, and taking away privileges. Many problem behaviours are maintained by social attention and positive attention can be a powerful consequence for strengthening appropriate behaviour.

The praise and ignore or pivot tool illustrate a positive change approach. Pivoting involves the use of attention as a reinforcer of appropriate behaviour. The pivot tool involves: 1) regularly and consistently attending to appropriate behaviour, while 2) ignoring harmless inappropriate behaviour. When inappropriate behaviour occurs, the strategy is to pivot attention away from the problem behaviour to another task or the appropriate behaviour of another individual, wait patiently for the targeted individual to engage in some appropriate behaviour, and then pivot attention and praise back to the individual (Whitman & Jackson, 2006).

If a desired appropriate behaviour is occurring too infrequently to provide sufficient opportunities for reinforcement, the reinforced practice tool can be used. This tool is especially appropriate for addressing tasks that the PWS individual may have been consistently resistant to completing. The steps for this tool are: 1) decide what the individual is expected to do, 2) provide opportunities to learn and practise the behaviour repeatedly with reinforcement, and 3) reinforce the behaviour when it occurs in a real situation. These steps should be repeatedly practised with reinforcement provided every time the sequence is appropriately performed. Once the behaviour has occurred a few times, it may not be necessary to provide praise at each occurrence. If the behaviour deteriorates unacceptably at a later time, the parent simply repeats the practice tool (Whitman & Jackson, 2006).

Another tool for making use of the reinforcers discussed above is the behaviour contract. This is a written agreement based on which an individual will earn a desired item or privilege based on the occurrence of a specifically defined behaviour. Behaviour contracts are usually used to address persistent problems, to establish new responsibilities, or to improve performance when the individual's responses have been inconsistent. The contract should also specify a daily consequence for completing tasks (e.g. sticker on a wall chart) and a long-term consequence (e.g. a trip to a pet store) for consistent completion of tasks (e.g. 4 out of 5 weekdays). Once the behaviour is occurring consistently, the tangible reinforcer can be used to address another behaviour, and the original behaviour can be maintained by the occasional praise and more natural consequences (Whitman & Jackson, 2006).



Whitman and Jackson (2006) suggest that dangerous behaviour or behaviour that is too disruptive to ignore, can be addressed with the stop-redirect-reinforce technique. This tool requires 1) immediate termination of the behaviour by interrupting it verbally and physically; 2) verbal redirection to an appropriate behaviour; and 3) immediate provision of positive attention when the redirected behaviour occurs. For behaviours that have not responded well to other attempts at change, or when the process of stop-redirect-reinforce has been unsuccessful, time-out from positive reinforcers may be used. Time-out is a specific detailed protocol that, when correctly employed, can be part of a very effective plan for eliminating serious, problem behaviour. Time-out should only be used with an individual who can be controlled physically by those expected to implement the time-out. Time-out will not be effective if improperly implemented. When first used, it often produces a burst of resistive or aggressive behaviour, which may be so severe that the intervention cannot be useful (Whitman & Jackson, 2006).

- **Medication for behaviour modification**

According to Whitman and Jackson (2006) the use of psychotropic and neuroleptic medications (designed to alter behaviour, mood, or thought by targeted action on brain chemistry) is increasingly advocated to supplement behaviour management strategies or as a primary intervention with severe emotional or cognitive disruption (e.g. depression, psychosis). The use of these medications in the PWS population remains one of the more controversial management strategies, due, in part, to a paucity of research in this area. Widely differing, often contradictory experiences with similar medications and dosages result in vastly differing clinical opinions regarding medication use in general and regarding specific medication.

An additional source of controversy is that medication is often used as a substitute for good behaviour management particularly for difficult and challenging behaviours, and in community living settings. This complicates decisions regarding medication use in this population. A range of effective medications is currently available; however, some patients respond well to one medication and not another. Available data suggests some cautions to consider when prescribing for this population. The cautions are physiological and behavioural. Some medication side-effects include

increased appetite, while others suppress cognitive abilities (Whitman & Jackson, 2006).

Due to an altered body composition and metabolism, individuals with PWS often have extremely idiosyncratic, frequently unpredictable, and often negative responses to medications. Many demonstrate much lower dose thresholds and tolerances and longer drug half-lives, than those of a non-affected population. Symptom response may be evident much sooner. For example, it is not unusual to observe improvement with selective serotonin reuptake inhibitor (SSRI) therapy in as little as 24 hours, particularly in the area of improved flexibility and reduced irritability. Finally, side effects may be different, more intense, and longer lasting in PWS individuals (Whitman & Jackson, 2006).

As described, many behaviours are commonly present in the behavioural phenotype of persons with PWS behaviours which often become exaggerated in frequency or severity with stress or physical illness. Many parents report feeling pressured to medicate their child when behaviours become exaggerated in response to stress, rather than altering the environment in order to manage stress. Many parents also have mentioned that the need to medicate is for the convenience of the service provider (e.g. group home staff, teacher) rather than the needs of the person with PWS (Whitman & Jackson, 2006).

## **2.20 Inpatient crisis intervention for persons with PWS**

Gourash, Hanchette and Foster (2006) summarized their clinical experience in caring for individuals with PWS who were hospitalized for crisis intervention. They state that crisis in individuals with PWS involves deterioration in the level of functioning across medical, behavioural, and/or psychiatric domains, or in the individual's support system. A crisis for a PWS individual may be abrupt or, as often occurs, is the culmination of multiple contributory events. Because of the unique and complex problems associated with PWS, these situations may benefit from a specialized approach implemented by a multidisciplinary team experienced with the syndrome.

Medical crises in individuals with PWS stem primarily from the consequences of a disorder of satiety. Rarely, stomach rupture from overeating has occurred. Food-seeking behaviour, as stated previously, may include foraging for food in and out of the home and consumption of spoiled, raw, frozen, or otherwise inedible foods. Older children and adults may steal food or money or use their own money to buy additional food. They may call food delivery services, elope in search of a restaurant, or enter a stranger's home to seek food. They may pick and break locks or steal keys to enter a locked kitchen, and may display violent outbursts or aggression related to food acquisition or to attempts to set appropriate limits. Families often find themselves in a situation that is spiralling out of control. Law enforcement may become involved further complicating management (Gourash et al., 2006)

Individuals with PWS have been described as more vulnerable to stress. Cognitive deficits often diminish their ability to adapt to change. They are exceptionally dependent on others to provide structure and control of their environment. Limited coping mechanisms may result in extreme maladaptive behaviours. Presenting symptoms leading to hospitalization include: depression; suicide attempts; dangerous, aggressive, destructive, disruptive, and intolerable behaviours; delusional thinking; and hallucinations. Self-endangerment, self-mutilation, and rectal picking can all result in additional medical complications. Stress appears to be a contributing factor in the onset of major psychiatric disorders like adjustment disorders, anxiety disorders, mood disorders, psychosis, and impulsive control disorders like intermittent explosive disorder (Gourash et al., 2006).

### **2.20.1 A specialized therapeutic milieu**

Gourash et al. (2006) outline a specialized therapeutic milieu which is an essential element in inpatient crisis management, and which is dependent upon trained and experienced staff. The ideal inpatient treatment plan for a patient with PWS has six major components: a) milieu management; b) behavioural intervention; c) psychological therapies; d) psychotropic medication; e) family/staff intervention; and f) disposition/systems intervention.

*a) Milieu management consists of:*

- Rules of conduct: which are explained and posted next to the patient's bill of rights in the individual's room.
- A daily schedule of activities: this comprises of a premeditated time line of activities, including wake up, grooming, therapies, meal times, exercise, leisure, rest, and bedtime. It defines the flow of the day and establishes structure, consistency, and predictability. This timeline is best presented to the patient in concrete form; a wall chart may be supplemented by a written schedule which the patient carries with him or her throughout the day.
- Psychological food security: the goal is to maintain food security across all settings, in and out of hospital. Food security is achieved when food access is controlled to the extent that there is no doubt about when, what, and how much they will eat; there is no hope of receiving any more; and there is no disappointment due to false expectations.
- Mandatory supervised exercise: must be scheduled into the daily plan. Walking is by far the best aerobic exercise. Two different periods for exercise are scheduled in the day.
- The day stops here!: if the flow of the daily schedule is interrupted by a behavioural situation (refusal, shutdown, or outburst), the daily schedule stops at that point until the individual has regained motivation and behavioural control in order to return to programming. Individuals are encouraged to return to scheduled therapies, and if the therapies have ended, they are prescribed "make-up work" which must be accomplished before moving to the next activity. It is essential to delay meals until make-up work has been completed. Sometimes individuals have behavioural shutdowns that can last as long as a full day. Nourishment should not be withheld, and an alternative calorie source is provided even if the privilege to a meal time is lost.

*b) Behavioural interventions*

Non-contingent reinforcement is the delivery of reinforcers independent of an individual's response, and is a powerful tool for establishing rapport. Typical non-contingent reinforcers include talking to the individual and providing leisure activities. Contingent reinforcement is the delivery of reinforcers dependent upon the individual's response. Contingent reinforcement is a powerful tool for shaping appropriate behaviours. Extinction, selective attention, praise, and differential reinforcement of other behaviours are examples of contingent reinforcement (Gourash et al., 2006).

*c) Psychological therapies*

Throughout inpatient hospitalization, individual and group therapy sessions focus on the acceptance of and building of expectations for a continuation of the therapeutic milieu following discharge. An appreciation for intellectual level and learning style is essential, and adapting the daily plan to meet the individual's unique pattern of strengths and weakness can lead to better compliance during the hospital stay. Strategies for adaptation can be tested during the hospital stay and taught to the family or other caretakers before discharge (Gourash et al., 2006).

The utility and effectiveness of psychological interventions is based on an individual's verbal and intellectual abilities. High functioning individuals may benefit from all psychotherapeutic and behavioural modalities. Clinical experience suggests that young adults with PWS are able to realize that their syndrome may limit their potential for independence, and that many of the life goals they share with their peers and siblings may never be actualized. They need support as they grieve the loss of a "normal" life (Gourash et al., 2006). Lower functioning individuals with limited insight may require individual therapy with supportive and psycho-educational goals like minimizing stress, enhancing coping abilities, and improving participation and compliance with the inpatient programme. Strategies and modalities for relaxation (progressive muscle relaxation, deep breathing, and sometimes visual imaging), anger management, social problem solving, and social skills training may be prescribed, taught to the individual with repetition and drill, incorporated into their daily plan, and implemented with prompts, cues and supervision (Gourash et al., 2006).

*d) Psychotropic medication*

The use of psychotropic medication is determined by: the patient's response to behavioural and eco-environmental interventions; psychiatric diagnosis; the nature of targeted symptoms; and the severity of impairment.

*e) Family and group home staff involvement*

Preparation for discharge begins before admission. Family education is an intense process that continues throughout hospitalization. Training sessions are attended by parents, step-parents, and siblings, as well as all involved extended family members. The goal of the training is to teach the caretakers how to devise, within the limits of their own situation, a therapeutic milieu similar to the one shown to be effective during hospitalization. The plan mimics the programme by providing the same structure, expectations, consequences, and schedule. The hospital exercise programme to which the patient has become accustomed is duplicated or modified, as needed, for the home environment. Psycho-educational intervention is essential for relapse prevention. Eco-environmental interventions can be developed after a thorough review of predisposing, precipitating, and perpetuating factors. This information provides a basis for understanding the strengths and weaknesses in the system of care (Gourash et al., 2006).

*f) Disposition planning*

Disposition planning includes an examination of all available short-term and long-term resources and living arrangements. Residential treatment or group home placement is explored when appropriate especially in the case of patients with an exceptionally high need for structure and supervision. PWS individuals are rarely well managed in facilities that do not have prior experience with the syndrome or a commitment to develop a programme specifically suited for individuals with the syndrome (Gourash et al., 2006).

## **2.21 CONCLUSION**

This chapter highlighted the possibility of early diagnosis and the importance of knowledge about PWS. This makes it possible to advise parents and caregivers about the characteristics of the condition, behaviour problems, and the eating disorder. Management of PWS is focused on addressing the symptomatic

characteristics of the syndrome, and is age dependent. Thus far, early management of PWS has been shown to improve the quality of life of individuals. Individuals with PWS can benefit from facilitated independence by offering them vocational training and placement.

This review of the mainly American literature suggests that as with all chronic illnesses, establishing residential facilities for individuals with PWS and supporting the primary caregivers are essential for the effective management of individuals with PWS. Hence, deciding on the appropriateness and type of residential support is one of the most important considerations facing any person and/ or the family that needs to cope with PWS. Through the partnership between parents, professionals and providers, PWS individuals could have a safe, consistent and stable home.

## **CHAPTER 3**

### **RESEARCH METHODOLOGY**

#### **3.1 Introduction**

In this chapter, quantitative and qualitative research are briefly discussed and the rationale for choosing qualitative research for this study becomes evident. Approaches to selecting samples are outlined and purposive sampling described in detail, as it was the chosen method for selecting participants for this study. An overview of the methods of data collection used in qualitative research are discussed, although emphasis is placed on the semi-structured interview. The procedure of data analysis is clearly sketched out, and attention is given to ethical considerations.

#### **3.2 Quantitative and qualitative research methods**

##### **3.2.1 Quantitative research methods**

There are several ways of classifying research methods. However, in this chapter attention is given to quantitative and qualitative research methods. According to Maree and Pietersen (2014) and Struwig and Stead (2004) quantitative research is systematic and objective, and involves large representative samples of the population under study. It uses these samples to generalize the findings to entire populations. The primary role of quantitative research is to test hypotheses.

Maree and Pietersen (2014) described two main categories of quantitative research designs: experimental and non-experimental. Experimental designs have been developed to answer a specific kind of question – the cause-and-effect question. It has three characteristics: 1) manipulation, where some of the participants receive some kind of treatment; 2) control, in which some participants are used as a control by not receiving the treatment; and 3) randomization, which is used to assign the participants to different groups (Maree & Pietersen, 2014).

In the experimental design all the participants are subjected to a pre-test and thereafter only the experimental group gets the treatment and then both groups do the post-test. The answer to whether the treatment was effective is obtained when comparing the scores of the post-test. Data usually come from a variety of sources



and are captured on different scales with each one based on the amount of information or characteristics of the information in the data. The names of these four scales are: nominal, ordinal, interval and ratio. Nominal carries the least information, ordinal carries slightly more, the interval even more, and ratio carries the most information (Maree & Pietersen, 2014).

Non-experimental designs are mainly used in descriptive studies, in which the sample which has been selected to take part in the research is measured on all the relevant variables at a specific time. No manipulation takes place. The most widely used non-experimental research design is surveys (Maree & Pietersen, 2014).

### **3.2.2 Qualitative research methods**

Qualitative research is a method of naturalistic enquiry which is usually less intrusive than quantitative methods and does not manipulate a research setting. This means that it aims to study people in their natural settings and to collect data as they occur. The research aims to understand the individual's view, without making any value judgments during data collection. The focus is on the meaning that participants in the study attach to their social world (Bowling, 2014).

Hence, emphasis is placed on their lived experiences. This gives perspective to the meanings that people place on events, processes, structures of their lives, and their perceptions, assumptions, prejudgments and presuppositions. This highlights the importance of getting close to those we study, attempting to see the world through participants' eyes, and conveying the experience in a way which is faithful to their everyday life (Ellis, 2004; Miles & Huberman, 1994).

Maree and Pietersen (2014) concluded that qualitative research methods are concerned with understanding the processes and the social and cultural contexts which influence certain behavioural patterns we observe in the participants we study. The research is mostly concerned with exploring the "why" questions. It aims to understand the meaning of human action and asks open-ended questions about phenomena as they occur in context rather than setting out to test predetermined hypotheses. Qualitative research is widely used in conducting psychological research (Carter & Little, 2007; Denzin & Lincoln, 2005).

Carter and Little (2007) wrote about analysing the qualitative data once it is gathered. They state that qualitative research means social research in which the researcher relies on text data rather than numerical data, and analyses the data in their textual form rather than converting it to numbers for analysis.

Ellis (2004) and Llewelyn and Kennedy (2003) further highlighted qualitative methods as being the more general and inclusive term. The term refers to a variety of research techniques and procedures. Approaches that could be considered qualitative include idiographic studies, ethnography, ethnomethodology, grounded theory, protocol analysis, discourse analysis, conversational analysis, constructivist approaches, humanistic approaches, phenomenology, hermeneutic investigation, conceptual studies, historical research, action research, and case study research (Maree & Pietersen, 2014; Stiles, 1993).

For the purpose of this study, case studies are used. The case studies incorporate ethnography, elements of auto-ethnography, and participatory action research.

### **3.3 Case study research**

According to Hyette, Kenny and Dickson-Swift (2014) case study research is an increasingly popular approach among researchers. It is defined as an investigation and analysis of single or collective cases, which are intended to capture the complexity of the object of study. It is defined by interest in individual cases, rather than the methods of inquiry used.

Hyette et al. (2014) proposed three types of case study design frameworks: the intrinsic case study; the instrumental case study; and the collective instrumental case study. The intrinsic case study is used to understand the particulars of a single case, rather than what is represented. An instrumental case study provides insight on an issue or is used to refine theory; the case is selected to advance understanding of the object of interest. A collective case study refers to an instrumental case which is studied as multiple, nested cases, which are observed in union, parallel or sequential order. More than one case can be simultaneously studied; however, each case study is a concentrated, single enquiry, and is studied holistically in its own entirety.

Therefore, for the purposes of this study, the collective instrumental case study was chosen. The case studies are presented in chapters four to eight.

Simon (2009) emphasized that the primary purpose of a case study is to generate in-depth understanding of a specific topic as in a thesis. Bowling (2014) further elaborates and highlights that the number of case studies used is usually small as the cases are intensively explored in-depth, retrospectively, and sometimes over time, through for example, detailed observations, interviews, and by obtaining information from records.

A case study is flexible, is neither time-dependent nor constrained by methodology, and has the potential to involve participants in the research process. It also provides an opportunity for researchers to take a self-reflexive approach to understanding the case and themselves (Simon, 2009).

Subjective data is an integral part of the case. It is through analysis and interpretation of how people think, feel and act that many of the insights and understanding of the case are gained. A case study acknowledges that the researcher is the main instrument in data gathering, interpretation and reporting (Hyette et al., 2014). Hence, I collected all the data and then analysed all the data.

Stake in Simon (2009) in outlining his view of a case study indicates that he draws from naturalistic, holistic, ethnographic, phenomenological and biographic research methods. May (2011) stated that case study research resonates with participant observation and ethnographic traditions. Therefore, in this study, elements of ethnography, auto-ethnography and participatory action research are incorporated in the case studies, in an attempt to generate a comprehensive understanding of them. This corresponds to what Bowling (2014) suggested that multiple research methods can be employed to fully investigate complex situations and to validate the findings.

### **3.4 Ethnography**

The word “ethnography” comes from two Greek words *ethnos* which means people and *graphein* which means to write, and implies writing about people (Maree & Pietersen, 2014). Genzok (1999) and Pelto (2013) state that ethnography is the

branch of anthropology that deals with a written, systematic description of a specific group or human culture.

Doing ethnography means using multiple methods of data collection like observation; interviews; collection of documents, pictures and audio-visual materials; and representations of artefacts. The main difference from other methods is that researchers do field work and collect their own data through their physical presence (Silverman, 2011).

Pelto (2013) further outlined and explained types of data collection that field researchers in ethnography undertake:

- The central core of ethnographic data consists of written notes and perhaps some audio recordings of individual interviews, plus field notes from observations of events, actions, and related descriptive information.
- A daily log of activities, which contains: (i) the amount of time spent on activities like interviews and direct observations and other brief notes about activities of the day; (ii) a personal diary of actions and thoughts related to field work including parts about planning, reviews of mistakes, emotional reactions to sensitive interviews, hunches and hypotheses about the field situation and the state of the data, and also other personal thoughts.
- Collection of secondary materials, including newspaper accounts, newsletters, local writing, and organization records.
- Structured survey forms, which are often a basic house-listing and demographic data set for describing the study of communities.
- Photographs, videos, and other visual documentation.
- Letters which field researchers write to families, colleagues, and friends, and which often have valuable syntheses of field information.

Conducting interviews and writing notes on my observations were the main methods of data collection in this study; in addition, photos were also taken.

Genzuck (1999) emphasized that ethnography relies on personal experience and possible participation, and not just observation. Ethnographers argue that it is

necessary to learn the culture of the group being studied, before providing explanations of the behaviour of its members. Ethnography refers to social research that has most of the following features:

- People's behaviour is studied in an everyday context, rather than under experimental conditions created by the researcher.
- Data are gathered from a range of sources but observation and/or relatively informal conversations are usually the main ones.
- The approach to data collection is unstructured in the sense that it does not follow a detailed, pre-set plan.
- The focus is usually on a single setting or group of a relatively small scale. The focus may even be on a single individual.
- The analysis of data involves interpretation of the meanings and functions of human actions and mainly takes the form of verbal descriptions and explanations with quantification and statistical analysis mostly playing a subordinate role, at most.

Ethnographers, when writing an ethnography, try to capture a segment of time in the lives of those they are observing, and describe it in a way that allows others to understand what is happening (Berger, 2001). Denscombe (2010) states that another characteristic feature is the role of the researcher in the research process. The researcher's identity, values and beliefs become submerged in the process and cannot be ignored, as they influence the findings. Ethnographic research therefore requires a degree of introspection.

### **3.5 Auto-ethnography**

Auto-ethnography is understood as being the process by which researchers choose to make explicit use of their own involvement and experiences, as an integral part of ethnographic research (Butz & Besio, 2004). In auto-ethnography, the experiences of the researcher are important for the purpose of extending sociological understanding. Research starts with the researcher's own experience as has been evident in the motivation for doing this particular study. In research that seeks to discover personal experience, there is a unique relationship between the researcher and the participants of the study, which cannot be ignored. It is suggested that the

freedom of the researcher to voice his/her experiences as a contributor in the research process and merging their experiences with those of participants, is needed to promote inquiry and knowledge advancement. If the researcher's experiences are overlooked, the writing is reduced to a summary and interpretation of others' work with nothing new added (Wall, 2006). The use of auto-ethnography alongside other well-known qualitative research methods, is suggested (Wall, 2006). Hence, in this study it is used together with elements of ethnography, and participatory action research in the case studies.

According to Ellis (1999) in auto-ethnography the researcher might integrate parts of his/her experience into each participant's story each of which can form a separate chapter. In each chapter, the researcher may reflect on different experiences they have had in each interview. This can be done by writing down their thoughts as they listen to the interview. The collaborative process allows readers to be more active participants in what they read, and also to witness the interaction between the researcher and the participant (Berger, 2001). In this study, this was achieved through reporting on my personal reflections and experiences, while conducting interviews and implementing interventions.

Auto-ethnography requires the reader to care, to feel, to empathize, and to do something. It needs the researcher to be vulnerable and intimate (Ellis & Brochner, 2006). In line with the above, it was crucial for me to congruently reflect on my own experiences during my interaction with all the participants, and also to report on the impact it had on me. This reflective process is described in detail in all the case studies.

### **3.6 Participatory action research**

Participatory action research (PAR) is built upon the action research model particularly incorporating an understanding of the power of group dynamics and relationships between individuals, groups and communities. PAR follows the process of planning, taking action, observing, and reflecting (Gray, 2014).

According to O'Brien (1998) action research also known as participatory research and is learning by going where a group of people identify a problem, doing

something to resolve it, seeing how successful their efforts were, and, if not satisfied try again. Therefore, this method was appropriate for this research as I went to the homes to assess the living environment of PWS individuals to make use of different interventions, and to identify the most effective interventions in the treatment of PWS participants and their primary caregivers. Where necessary, an individualised programme or intervention was developed and the participants were fully involved in the whole process.

According to Bergold and Thomas (2012), in participatory research the willingness on the part of the participants to disclose their personal views of the situation and their own opinions and experiences, are required. Participatory research is conducted directly with the immediately affected persons; the aim is the reconstruction of their knowledge and ability, in a process of understanding and empowerment. Participatory action research recognizes that knowledge is the meaning people attribute to their experiences. Hence, I focused on these experiences, in order to implement different interventions and to develop programmes or interventions, as appropriate, to individual experiences as stated above.

### **3.7 Participants and setting**

Participants were individuals diagnosed with PWS, and also their primary caregivers. Simon (2009) uses the word “participant” to signal her preferred way of engaging people in the study in a shared experience which they can value. A primary caregiver in this study was identified as the person who took responsibility for the PWS individual, ensuring that their needs were met, and they stayed with them on a daily basis.

The participants were members of the Prader-Willi Association of South Africa (PWSSA). Permission was requested from PWSSA prior to conducting the research. The reason for choosing this population was because they are currently a minority group and are not widely available. Due to the limited available population of individuals with PWS, only five individuals diagnosed with PWS and their primary caregivers were chosen to participate in the study. This decision was supported by Lewis (1999) and Neuman (2003) in their views that qualitative studies are more in-

depth than quantitative studies and therefore make use of a smaller number of participants.

An appointment was made with the participants for the time and place that was convenient for them. Pseudonyms were used to identify the participants and primary caregivers. Approximately two follow-up appointments where interventions were implemented were conducted in the individual's home.

### **3.8 Inclusion and exclusion criteria**

The study included individuals who had been diagnosed with PWS and excluded those who were suspected to have the condition.

Only primary caregiver/s who took responsibility for the individual, ensuring that their needs were met and who stayed with them on a daily basis, were included. The primary caregivers in this study were all parents.

The nature of PWS and the family dynamics and problems at schools will require full-on and repeated engagement during the intervention implementing process. This considered, only participants who resided in Pretoria and surrounding areas, and who could speak English and Sesotho were included in the study as I can speak these languages.

### **3.9 Sampling**

Sampling procedures for qualitative research differ from quantitative research, in that random selection and generalization are not a primary consideration in qualitative research. Qualitative research focuses on the depth of the data and therefore qualitative researchers select samples purposefully, rather than randomly (Struwig & Stead, 2004).

According to Denscombe (2010) the basic principle of sampling is that it is possible to produce accurate findings without the need to collect data from every member of the population. Two types of sampling methods are used in research to select samples: probability sampling or non-probability sampling. These are now discussed:



### **3.9.1 Probability sampling**

Probability sampling relies on random selection. It is based on the theory that the best way to get a representative sample is to ensure that the researcher has no influence on the selection of participants to be included in the sample. The sample is selected randomly from the population being studied. Probability sampling is best suited for a large population, and is often associated with quantitative data (Denscombe, 2010).

There are several different types of probability sampling:

#### **3.9.1.1 Systematic sampling**

Systematic sampling adheres to the principle of random selection: the researcher selects participants in a systematic way for example, picking every tenth participant from the population (Denscombe, 2010). This sampling method is simpler and quicker than the use of random numbers. On the other hand, this method also has constraints. It relies on the availability of a complete, unbiased population list (Bless & Higson-Smith, 2000).

#### **3.9.1.2 Cluster sampling or multi-stage sampling**

Cluster sampling is done by dividing the population into large clusters or groups and then probability samples are selected from the groups. The selection process uses random sampling; it also involves selecting samples in stages (Denscombe, 2010). The first stage is to start by sampling the population which is more general than the final one. In the second stage, based on the first sample, a new population is considered, which is less general than the first one and a new sample is subsequently determined. The procedure is continued until the population to be investigated is reached, and a final sample is drawn (Bless & Higson-Smith, 2000).

#### **3.9.1.3 Stratified sampling**

Stratified sampling subdivides the research population into different subgroups called strata, and then chooses the required number of participants from each stratum (Bless & Higson-Smith, 2000; Denscombe, 2010).

### **3.9.2 Non-probability sampling**

In non-probability sampling, the probability of any particular member of the population being chosen is unknown (Struwig & Stead, 2004). Bless and Higson-Smith (2000) and Bowling (2014) have stated that non-probability sampling is widely used with qualitative data where the sample is chosen by the researcher based on the following:

- It is impossible to include a large number of participants.
- The researcher does not have sufficient information about the population.
- The researcher may not know how many people make up the population.

The types of non-probability sampling are:

#### **3.9.2.1 Convenient or accidental or availability sampling**

This method refers to situations when population elements are selected based on the fact that they are easily and conveniently available. It consists of taking all cases available until the desired sample size is reached. The approach is usually quick and cheap, but does not result in representative samples (Bless & Higson-Smith, 2000; Maree & Pietersen, 2014).

#### **3.9.2.2 Quota sampling**

The researcher has to first identify categories of people that need to be in the sample, and also the required number in these categories (Maree & Pietersen, 2014). Participants may be selected based on their characteristics such as age, income, socio-economic status, and gender. The participant must comply with certain criteria before being included in the sample (Struwig & Stead, 2004).

#### **3.9.2.3 Snowball sampling**

This method is often used in cases where the population is difficult to find or where the research interest is in an interconnected group of people (Maree & Pietersen, 2014). It emerges through a process of reference, from one person to the next (Bowling, 2014).

#### **3.9.2.4 Purposive or judgemental sampling**

Qualitative research samples are chosen purposively. This means that they are selected to serve an investigative purpose rather than to be statistically representative of a population (Carter & Little, 2007).

In case studies where the aim is to understand or gain insight into the case, purposive sampling is often used (Simon, 2009). Bless and Higson-Smith (2000) acknowledged that this sampling method is based on the judgement of the researcher regarding the characteristics of a representative sample. A sample is chosen based on what the researcher considers to be typical units. The strategy is to select units which are judged to be the most common in the population being investigated. The danger of this type of sampling is that it relies more heavily on the subjective considerations of the researcher than on objective criteria. The above factors were all considered and for the purposes of this research study, purposive or judgemental sampling was used.

Most qualitative studies use purposive sampling in terms of the selection of a smaller number of participants (Gacomini & Cook, in Sethuntsa, 2009). Peltó (2013) adds that most ethnographic samples are opportunistic or purposive. Purposive sampling aims to sample small groups of people or settings with particular characteristics. The advantages of sampling smaller numbers over complete population coverage, are financial and a better quality of data are also obtained (Bowling, 2014). As stated earlier, only five individuals diagnosed with PWS and their primary caregivers were chosen to participate in this study. This approach is supported (as mentioned above) by various authors in their views on purposive sampling.

### **3.10 Methods of data collection**

Qualitative data collection methods include:

#### **3.10.1 Documents**

Documents focus on all types of written communication that may shed light on the phenomenon being investigated. Written data sources may include published and unpublished documents, reports, e-mail messages, faxes, and newspaper articles (Carter & Little, 2007; Maree & Pietersen, 2014).

### **3.10.2 Observation**

Observation is a process of recording the behavioural patterns of participants without necessarily questioning or communicating with them. It is used to enable the researcher to gain a deeper insight into and understanding of the phenomenon being observed. It allows the researcher to hear, see, and begin to experience the participant's reality. Before the researcher uses observation as a data gathering method, they must, however, know exactly what to observe (Maree & Pietersen, 2014). During the interviews and intervention process I observed the non-verbal behaviour of the participants. I worked closely with the participants with the aim to develop effective interventions and hence the decision to incorporate participatory action research into this study. This implies that I was a participant observer.

### **3.10.3 Interviews**

There are many possible ways of gathering information directly from participants and the first of these methods is the interview (Bless & Higson-Smith, 2000). An interview is a verbal exchange in which the interviewer attempts to acquire information and has an understanding of the interviewee (Gray, 2014).

By using interviews, the researcher can reach areas of reality that would otherwise remain inaccessible such as people's experiences, opinions, values, aspirations, feelings, and attitudes (Denzin & Lincoln, 2005; May, 2011).

There are different types of interviews:

#### **3.10.3.1 Structured interviews**

Structured interviews are often used in surveys, and are based on the same idea as questionnaires. They have a standardized way of asking questions. The interviews enable comparison of answers across participants and ensure that data analysis is relatively easy (Brinkmann, 2012). The structured interview does not allow the interviewer to probe for further information which is not covered in the interview (Struwig & Stead, 2004).

### **3.10.3.2 Unstructured or open-ended interviews**

Unstructured interviews place emphasis on interviewee thoughts. The researcher's role is to be as unintrusive as possible by introducing a theme or topic and allowing interviewees to develop their ideas and to freely express themselves (Denscombe, 2012). Struwig and Stead (2004) mentioned that the interviewer must refrain from imposing their viewpoint on the participants, but must listen to participant views. It is important for the interviewer to establish rapport with participants.

Bowling (2014) advocates using unstructured interviews. He described the unstructured interview as a face-to-face interview using an interview schedule with the topics listed, with few specific questions, and with no fixed questions. These interviews aim to be in-depth.

### **3.10.3.3 Group interviews and focus interviews**

Group interviews and focus interviews include more than one participant at a time, and the researcher can include a large number of participants in the research process. They constitute a valuable tool of investigation, allowing researchers to explore group norms and dynamics around issues and topics which they wish to investigate. Participants are encouraged to talk to one another, to comment, discuss their opinions and to answer questions without waiting for guidance from the interviewer (Denscombe, 2012).

### **3.10.3.4 Semi-structured interviews**

Semi-structured interviews are one of the most widely used interview techniques in qualitative research in which the researcher knows in advance what specific aspects of an experience they wish to have the subject cover in their discussion (Judd, Smith, & Kidder, 1991). Semi-structured interviews are flexible regarding the order in which topics are covered, and the answers are open-ended. This allows participants to answer according to their own terms, and they are not limited by standardized interview permits (May, 2011).

In this study, data were collected using semi-structured interviews. Interviews were conducted with the primary caregiver and the individual diagnosed with PWS. I had a

checklist of topics as a tool rather than fixed questions because there was important information that I needed from all participants (see Appendix IV).

I also had a brief, structured list of questions about the participants' biographical details which was the structured section in my semi-structured interview. These questions related to: age, geographical area, ethnicity, socio-economic status, gender, date of birth, occupation, education, date, place, time, religion, marital status, and number of dependants (see Appendix IV).

This method is supported by Bowling (2014), who stated that when investigators require more specific information, a semi-structured format is used. With this method the interviewer guides the interview on the topic of interest by asking specific, open-ended questions. The interview is, however, still carried out in-depth.

### **3.11 Procedure**

A permission letter was sent to the PWSSA (See Appendix I). I then presented my proposal at the PWSSA Annual General Meeting (AGM). Permission to conduct the study was resolved with the families who attended the AGM. A letter was sent to the Association when it was time for data gathering.

An appointment was made with the primary caregiver at a place convenient for them to be interviewed. Interviews were conducted with the primary caregiver and with individuals diagnosed with PWS. I conducted the interview on the date set for the appointment. A voice recorder was used to record the interview, but only with the participant's permission. A voice recorder was used to ensure that no data were lost. Voice recorders and other electronic recording devices can be an important aid to effective communication. Many ethnographers consider that practically all structured and semi-structured interviews should be recorded with audio equipment (Pelto, 2013).

According to Simon (2009) audio recording has several advantages. First, it ensures accuracy of reporting; secondly, it frees you from having to write everything down so that you can concentrate on the interview process and respond fully to the interviewee; and thirdly, you can compare data obtained from the recording with data

obtained from taking notes. The interviews were conducted in English and Sesotho. Interviews in Sesotho were transcribed and then translated to English.

Observations were documented by writing notes. Observation of behaviours is a tool for understanding more than what people actually say about situations and can help to comprehend complex situations more fully (Bowling, 2014).

Data on physical and behavioural characteristics were collected through my observations and secondary observations of the individuals with PWS made by their primary caregivers. Therapeutic sessions were also recorded using the voice recorder. Detailed notes on the therapeutic process were documented and interviews were transcribed verbatim.

Follow-up appointments were made in order to apply different interventions. Financial implications were involved, including transportation costs, printing costs and editing costs which I funded.

### **3.12 Method of data analysis: Thematic analysis**

There are many ways to analyse information about participant experiences, and thematic analysis is one such way (Aronson, 1994; Boyatzis, 1998; Bryne, 2001). According to Braun and Clarke (2006) thematic analysis is a method for identifying, analysing, and reporting themes in the data. It is a widely used form of analysis, but is rarely acknowledged and there is no clear agreement about what it is and how to go about doing it. However, thematic analysis is used to explore and report experiences, meanings and the realities of participants. It is a method that works to reflect reality and to reveal the underlying meanings. Data usually originate from interview transcripts or observational notes, and must represent major themes or categories that describe the phenomenon being studied (Bryne, 2001; Fereday & Muir-Cochrane, 2006).

Simon (2009) emphasized that simply presenting quotations from interviews or observations without any thematic structure, analysis or interpretation, is unlikely to convey the meaning of the case.

All interviews were transcribed and translated to English. The process of transcription was time consuming. However, it was also a valuable part of the research, because I was familiarized with the data (Denscombe, 2010).

The transcripts were analysed using thematic analysis. I personally did the analysis for each case study, in order to preserve all the data gathered. The case studies were presented in different chapters.

Different authors and researchers use varying steps to conduct thematic analyses. The version used by Sethuntsa (2009) was used for the purpose of this study. This is where the researcher uses a combination of steps, from different authors, to analyse data. Silverman (2011) supports this approach by stating that with qualitative data analysis, there are no specific rules or formulae for analysing data, except guidelines and procedural suggestions which depend on the researcher's judgment and creativity.

According to Bowling (2014) in order to analyse and present qualitative data, the investigator must be thoroughly familiar with the field notes, the tape recordings, their transcripts, and any other data collected.

The seven steps used throughout the analysis were as follows, and were applied to all transcripts of the participants:

*First step:* I familiarized myself with the data by reading and re-reading the transcripts (Braun & Clarke, 2006).

*Second step:* I engaged with the data by reflecting on what each participant communicated, their thoughts, comments, the interview process, and the emotions presented (Braun & Clarke, 2006; Lyons, 2007). Notes were made on the transcripts in respect of anything that appeared to be significant and of interest (Lyons, 2007).

*Third step:* I summarized the brief biographical information of each participant and then identified natural meaning units in the interview data. Frith and Gleeson (2004) and Kruger (1979) defined meaning units as statements made by the subject which



are self-definable and self-delimiting in the expression of a single, recognized aspect of the subject's experience.

The data were analysed and presented in relation to the topics that form the framework for the semi-structured interviews (Fereday & Muir-Cochrane, 2006; Frith & Gleeson, 2004). The natural meaning units were supported by quotes from the text to ensure that data interpretations were directly linked to the words of participants. According to Fereday and Muir-Cochrane (2006) participant reflections, conveyed in their own words, strengthen the validity and credibility of the research.

*Fourth step:* based on the previous steps, I presented a description of each participant's experiences related to the various questions posed during the interview. I identified possible underlying meanings in the text and I later added my observations regarding the whole interview and therapeutic process.

*Fifth step:* I identified themes from the data of participants in relation to each topic. A theme captured something important about the data in relation to the research question, and represented some level of patterned response or meaning within the data set. The researcher's judgement is necessary to determine what a theme actually is (Braun & Clarke, 2006; Ryan & Bernard, 2000).

Part of the flexibility of thematic analysis is that it allows the determination of themes in several ways (Braun & Clarke, 2006). A theme in this study was an experience that had been very strongly emphasized by one participant as well as those experiences mentioned by more than one participant.

*Sixth step:* I presented an overall description of the identified themes that emerged.

*Seventh step:* I integrated the findings with the literature reviewed in chapters one and two, and the findings of the therapeutic process (Aronson, 1994; Boyatzis, 1998; Bryne, 2001). Due to the fact that the case studies will be presented as stand-alone in separate chapters, the supporting literature may be repeated in each case study. This is done to ensure that the reader does not have to refer to other chapters to understand the discussion. The step-by step process of analysis outlined above is a

method of demonstrating transparency in relation to how data was analysed (Fereday & Muir-Cochrane, 2006).

### **3.13 Validity and reliability**

Validity and reliability concern trustworthiness. Reliability refers to the trustworthiness of observation or data. Reliability is a matter of whether a particular technique applied repeatedly to the same subject yields the same results each time. Validity refers to the trustworthiness of interpretations or conclusions. Validity also refers to the extent to which an empirical measure adequately reflects the real meaning of the concept under consideration (Llewelyn & Kennedy, 2003).

The following measures were implemented to ensure reliability and validity in the study:

- The interviews were recorded to guarantee that information was not lost or misinterpreted.
- The data collection procedure was documented in detail.
- Data analysis steps were based on published guidelines which I had previously used.
- Research supervisors gave guidance on the process of data collection and data analysis.

### **3.14 Ethical considerations**

According to Gray (2014) and Struwig and Stead (2004) research ethics refer to the moral principles guiding research. These are based on conducting research in a way that goes beyond merely adopting the most appropriate research methodology and involves conducting research in a responsible and morally defensible way. Such guidelines seek to prevent researchers from engaging in scientific misconduct such as: distorting and inventing data, plagiarizing the work of others, republishing their data as an original contribution without proper acknowledgement, deceiving people, and falsely reporting results.

Denscombe (2010) stated that there are certain measures which researchers are expected to put in place in order to minimize the risk of harm to participants:

- Participants should remain anonymous.
- Data should be treated as confidential.
- Participants are informed and understand the nature of the research and their involvement.
- Participants voluntarily consent to being part of the research.
- The use of deception is avoided.
- Researchers must act professionally and with integrity.

For the purpose of this study I obtained approval from the chief executive officer of the Dr. George Mukhari Academic Hospital and from the Manager of the Clinical Psychology Unit where the research was conducted. Permission to conduct the study with registered members of the Prader-Willi Syndrome Association of South Africa was obtained from the Association and from its members (Appendix I and II). Ethics clearance was obtained from the Department of Psychology's Ethics Committee of the University of South Africa prior to conducting the research.

Prior to conducting the research, participants were given verbal information regarding the purpose and nature of the research project. Participants were provided with adequate information regarding the research project so that they could make an informed decision regarding their involvement. This was achieved by giving a brief presentation at the PWS Association's yearly information session.

Prior to collecting data, participants were informed that all interviews would be voice recorded. Participants were informed that they were free to participate or decline to participate, and it was emphasized that participation was voluntary and that they had a right to withdraw at any time. All participants gave written consent (HPCSA, 2002) (see Appendix III). Participants were assured that information would be treated confidentially (Bless & Higson-Smith, 2000).

### **3.15 Conclusion**

In this chapter the rationale for choosing a qualitative research method for the current study was made clear. The sampling method was influenced by the fact that the population being studied is limited and not easily accessible. It was not viable to

get a representative sample. The main aim was to gather adequate information from the participants, in order to get an in-depth understanding of their experiences in order to intervene effectively.

It is obvious that the researcher's experiences during the research process cannot be excluded, as they form a significant part and have to be included when making sense of the data. There are many methods of data collection; however, semi-structured interviews were the most appropriate method for this study, to ensure that adequate information was gathered.

There are no fixed methods or numbers of steps for analysing qualitative data. Hence, thematic analysis was used in this research, because it is best suited to situations where it can be used to explore and report experiences, meanings, and the realities of participants.

There is no doubt that ethical considerations form the basis of the research process, as all participants have to be protected from harm, need to be treated with dignity, and the researcher must maintain professionalism.

## **CHAPTER 4**

### **WILNA'S STORY: "ACCEPT THE WAY I AM"**

#### **4.1 Introduction**

This chapter presents a case study of the lived experiences of the primary caregiver, Rina, and Wilna, an individual diagnosed with PWS. The challenges they encountered are expanded on. This case study demonstrates that caring for an individual with PWS is time consuming for the primary caregiver, and living with PWS has psychological impact. The findings are presented based on topics discussed in the interview and the themes that emerged. The statements are supported by quotes from the transcripts, which are italicised and indented.

I conducted two lengthy sessions of about two hours and thirty minutes each, with Wilna and Rina. In the first session Rina was interviewed and after her interview session, Wilna was interviewed. The first session focused on the interview questions and guidelines (see Appendix IV), while the second session was aimed at assessing the effectiveness of the interventions they were already implementing. An intervention was then formulated based on behaviour therapy, in order to manage Wilna's outbursts. A brief follow-up session was conducted to assess the effectiveness of the intervention. The first two interviews were transcribed and process notes were written for the follow-up sessions using the voice recorder to preserve all the data. The findings were then integrated with the literature.

#### **4.2 Family background**

This family comprises of the father, the mother and one child. They reside in a large and beautiful home in Muckleneuk, a secured suburb in Pretoria. The family income is more than R20 000 a month, and they own a car. They are Christians who attend the Dutch Reformed Church on a weekly basis. Their home language is Afrikaans but the interview was conducted in English, and they could express themselves fairly well in English. They are affiliated with the Prader-Willi Syndrome Association of South Africa (PWSSA).

### **4.3 The context**

I was very anxious when I started with data collection – it seemed to be a daunting and overwhelming process. It was not easy to initiate, and in order to gain the courage to begin with the research, I started with this particular family due to my previous interaction with Rina, who made me feel comfortable and put me at ease. The interview was conducted at the participants' home in Muckleneuk in Pretoria. I felt really welcomed by Rina (the mother and primary caregiver) and Wilna (the individual diagnosed with PWS) who further made it easy for me to engage with the family. The interview was first conducted with the primary caregiver alone, and Wilna joined us later.

Rina is a 70-year-old white married female. She holds a bachelor's degree in speech therapy and is a qualified speech therapist. She stopped working as a speech therapist at the end of 1982, just before Wilna was born. Since then she has been a house-wife, taking care of Wilna who was diagnosed with PWS, and participating in PWS-related activities. She was welcoming, friendly and warm. She spoke about her experiences in an emotive manner; however, when she became emotional she dismissed her emotions and was quick to regain composure.

Wilna is a 33-year-old, white single female. She has no children and is currently unemployed. In terms of her highest level of education, she attained a special education certificate. She presented as friendly and warm. She had observable dysmorphic features, gait difficulties, short stature, small feet, speech difficulties, and when she spoke the saliva thickened and filled her mouth. The interview with Wilna did not take long due to the speech difficulties, and because she did not elaborate much on some of her responses. Throughout the interview, she continuously snapped her fingers.

According to Lewis (2006) the speech and language skills of individuals with PWS are reported to be below expectations based on intellectual levels. The difficulties include poor speech-sound development which includes errors due to poor motor abilities associated with the production of speech-sounds and errors in applying linguistic rules to combine sounds to form words; and also reduced oral motor skills and language deficits. Language problems include deficits in vocabulary, grammar,

morphology, narrative abilities, and pragmatics. This explains why Wilna was struggling with narrating and formulating sentences. She mostly gave one-word answers and did not elaborate on her responses. She tried to elaborate when she spoke about her tantrums and the struggle for acceptance which suggests that she was passionate about the issue.

#### **4.4 Lived experiences**

The lived experiences of Rina and Wilna, were explored by conducting semi-structured interviews with them. From the transcribed interviews and sessions, certain themes were identified, which will be discussed in the following sections.

##### **4.4.1 Themes identified from interviews with Rina**

In the interview with Rina, the following themes became evident:

###### **4.4.1.1 Pre- and post-delivery**

Rina was 36 when she fell pregnant with Wilna. During her pregnancy she had decreased foetal movement which was worrying for her. She then asked her doctor about it, but the doctor explained that sometimes when the mother moves the baby also moves, and when the mother sits quietly the baby also stops moving. She mentioned that she is now informed that decreased foetal movement is one of the significant symptoms of PWS. According to the literature, this is called hypotonia and is prenatal in onset. It usually manifests amongst other characteristics, as decreased foetal movement (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

*“Yah! or a little something but no movement, no. And that’s one of the actually symptoms. Actually, you have to be worried about that.”*

Her pregnancy was full-term, but she had a caesarean section because the baby was not gaining weight. She gave birth in HF Verwoerd Hospital in Gauteng and was satisfied and impressed with the level of care she received. After delivery her daughter Wilna was floppy; she only gained muscle strength when she was two months old. She did not cry, even when she was hungry. She had to wake her up to feed her. Once again, due to hypotonia in infancy, there is decreased movement and

lethargy with decreased spontaneous arousal, weak crying, and poor reflexes including poor sucking that leads to early feeding difficulties and poor weight gain (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

*“You know Wilna was sooo floppy, and only at two months, you know, when I put her on the bed with the head this side [tilting her head to the left] it stays like that, and then at two months one day when I came into the room the head was to the other side [tilting her head to the right], then I realized she had turned.”*

*“Yah to feed her, and when she uh, uh was awake, yes and then she was, she didn’t cry for food. She didn’t cry.”*

McCandless et al. (2011) state that PWS should be considered in any infant with significant poor muscle tone (hypotonia), particularly in settings of poor feeding and reduced spontaneous arousal for feeding, as was observed in Wilna’s case.

#### **4.4.1.2 Feeding problems**

Difficulty in sucking is one of the commonest symptoms of new-borns with PWS, and therefore a feeding tube was inserted in the nose and Wilna was admitted to hospital for two months post-delivery. The feeding tube was changed every week. Following discharge, she had to go to the hospital on a weekly basis to have the feeding tube changed.

Wilna was fed through the feeding tube with Rina’s breast milk for three months and Rina expressed her struggles in doing this. This was in line with the literature that states that assisted feeding through a feeding tube and/or special nipples with increased feeding times, are necessary for a period of time, usually weeks to months (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

*“I left her in our room and I, and I fed her you know with a tube, and it took ages, and for the first three months she got breast milk.”*

She always had to wake her up every two hours to feed her as the infant would not wake up on her own. This is part of the sleeping problems documented for PWS infants. Cassidy and Driscoll (2009) and Honey (2010) stated that sleep problems



are commonly observed and reported them to be one of the characteristics in individuals with PWS.

*“I had to wake her up in the night, every two hours, she didn’t uhh, wake up for a feeding.”*

#### **4.4.1.3 Milestone developments**

During infancy it was noticed that Wilna could not sit and it took a long time for her to develop muscle strength which could be attributed to hypotonia. Subsequently, her milestone developments were delayed. She started walking at 25 months and her language development was also delayed. This concurs with Cassidy and Driscoll’s (2009) statement that there are delayed milestones, including gross motor and language delays. Early milestones are reached, on average at double the normal age for example, sitting at 12 months, walking at 24 months, and speaking words at 2 years.

*“Yah! Yah!, and but actually we experienced many problems with Wilna when she was a baby to get her strong enough. But once she started to walk that was about 25 months. She was kind of ok and she was always uhm, her milestones were delayed.”*

#### **4.4.1.4 Diagnosis**

Wilna was not diagnosed immediately after birth, but the doctors were speculating, and mentioned a few conditions, including PWS.

*“They wasn’t [sic] sure, they mentioned a few things and, uhm, Prader-Willi was one of them.”*

When Wilna was three years old, she was officially clinically diagnosed with PWS based on symptoms, but Rina was ambivalent about the diagnosis as she found it difficult to accept, but, on the other hand, it was a relief to eventually have a diagnosis.

*“Yah! yah! But I thought argh! But he said no, let’s rather leave it and then when Wilna was three years we went to Dr Otto, he was one for syndromes.”*  
*“And then he confirmed the diagnosis on symptoms.”*  
*“Then we had a name for this problem.”*

I was impressed to learn that 33 years ago doctors could identify the symptoms as relating to PWS especially given that when I currently speak to some medical doctors, paediatricians, and neurosurgeons, they don't know what PWS is.

#### **4.4.1.5 Medical problems**

Wilna's overall physical health is good, according to Rina. In early childhood she developed an ear infection, and that is when she realized that she did not get a fever even when she had an infection. She further mentioned that Wilna dresses based on the weather forecast, because she sweats easily. The literature confirms that some children with PWS may have temperature instability and/or insensitivity, and also a high pain threshold (Whittington & Holland, 2010).

*"Wilna is actually very healthy ... uhm as a baby she had, uhm, ear infections and I experienced there that she is actually quite ill without a fever."*

*"Wilna dresses according to the weather focus ... she sweats quickly, that's the problem, uhh [pause], but she is fine."*

#### **4.4.1.6 Treatment received**

As Wilna got older her muscle tone got stronger, but due to poor muscle tone as stated above all her milestones were delayed including speech. Subsequently in early childhood she attended speech therapy, physiotherapy, occupational therapy and also consulted with a dietician. In late childhood she was managed by the family doctor who started Wilna on sertraline in order to manage her stress levels.

Sertraline is a SSRI antidepressant (Sadock & Sadock, 2003) which is the class of antidepressants most commonly used for PW patients. They are useful, not just for depression, but also for other problems which affect PW patients such as obsessive-compulsive disorder, aggression, and impulsive behaviour (Boyle, 1997).

A paediatrician was also consulted, who prescribed glucophage as there were possible signs of diabetes. At the age of 17 to 18 she received growth hormone therapy for 18 months. I found it interesting that she started it so late, because McCandless et al. (2011) state that clinical experience suggests that growth hormone treatment can be beneficial for an individual with PWS as early as two to three months of age. If treatment is intended to increase height it needs to be initiated before the normal age of puberty.

At the age of 19 to 20, Rina took Wilna to a psychiatrist, but with time the consultations became too expensive and they took Wilna back to their family doctor who continued prescribing sertraline. Wilna was constantly in hospitals for most of her childhood, and Rina does not want to continue taking her to different doctors as she feels this practice can be overdone. Rina is content with just taking Wilna to the family doctor to manage her, as her overall health is currently good.

*“Yah, ok, but in any case, Wilna is healthy, em, and I just felt ... you can overdo it as well ... now we more or less, only when there is a problem, we go to a doctor.”*

#### **4.4.1.7 Impact of PWS on Rina**

Rina's life changed immediately after Wilna was born. It was an unexpected, overwhelming and sudden change. It was a daunting experience for her especially given that it was her first baby. She tried to read articles about the syndrome, but it was difficult to acquaint herself with information about PWS. Rina seems to have dedicated most of her adult life to her daughter and to PWS-related activities.

*“You know and, uh, it took me [pause], I can't tell you how long it took me to read that article, I started and I read a few spaces it just was too ... oo [stuttering].”*

*“That's why you know, my whole life is Prader-Willi ... but it is my life! It is my life!”*

*“Immediately! Yah! That's why I said, uhm, uhm, to the people who I addressed really, uhm ... in any case I said it changed, it has really changed.”*

#### **4.4.1.8 Schooling**

Wilna started pre-school at a nursery for children with intellectual disabilities, and was again enrolled in a primary school for children with intellectual disabilities until grade three. She was struggling to cope with her school work, and subsequently she repeated grade two. She was assisted with her school work every afternoon and on weekends. Regardless of the IQ score, most people with PWS have multiple severe learning disabilities and also poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

*“We worked very hard ... very hard every afternoon, Saturday, Sunday and then it was in August of standard three that she started to cry that specific evening.”*

The turning point for Rina was when Wilna came back from school, having seen grade four books, and she was crying, worried that she would not be able to cope with the work. Rina blamed herself and felt like a failure because her daughter was unable to cope with her school work, despite their efforts. She then made a decision to stop school and enrolled her in a pre-vocational school, where they focused on practical work and on her strengths. That is where she got a special education certificate. I was then wondering about where her husband was, as he seemed to be silent in the decision-making process.

*“Because I felt I was a failure, but I realized, I tried to get something out of this child ... that she didn’t really have, and what was going to be the difference with her having standard five, [or] having standard three.”*

At school, Wilna not only experienced problems with her school work, but also problems with other children. During break time other children would tease and bully her. This concurs with Whittington and Holland (2010) who reported that, by school going age, and in addition to intellectual disabilities, social difficulties become more apparent.

*“I think she had difficulties with the other children ... they teased her, but she managed.”*

#### **4.4.1.9 Food-related problems**

During early childhood Rina mentioned that Wilna did not have any food-related problems. Rina always ensures that, as a family they eat healthily, and has never introduced Wilna to snacks like chips and sweets. Instead, she would buy things for her like cocktail tomatoes, as a treat.

*“I never buy ... when she was little it was her treat to buy those small ... when I am in Woolworths I allow her to buy those little tomatoes – she is very fond of them ... but never ever chippies, sweets.”*

She observed that, as a child Wilna enjoyed pretend play where she owned a restaurant and was preparing food. Rina had reviewed the literature and had established satiety and other food-related behaviours were a significant problem for children with PWS. But this was not the case with Wilna. As a result, she did not implement measures to prevent Wilna from having unlimited access to food.

*“What she did, when she was about say three to five years, that stage she liked to play having a restaurant ... and that I also think she, she don’t, doesn’t have that hungry feeling. Wilna can tell you ‘that was enough’ or she will ask, it doesn’t happen often, but then she will ask you ‘can I have this tomorrow?’”  
“I became perhaps a little more relaxed ... and I was naive to think that she will never start stealing food ... and I didn’t lock anything.”*

At around 18 to 19 years Rina observed that Wilna was gaining weight and it was then that she discovered that Wilna had started stealing food. When she realized this it took her by surprise.

*“Yah! And, but then I also I didn’t and I wish I was more aware of that then.”*

For example, Wilna was in charge of making tea or coffee in the afternoon and used to steal rusks. At one point, Wilna used 10 sweeteners in a cup of coffee and drank seven cups of coffee a day – which made it 70 sweeteners a day. When she was confronted about this and the intake was reduced from ten to six, she had terrible tantrums. The other interesting point that Rina mentioned was that Wilna did not comprehend uneven numbers hence she then reduced the intake to six and then to four.

*“Because then suddenly I uhm, became aware that she uses 10 ... yah! and then we argh [sigh] really had terrible fights, she screamed and she was so cross with me. Then we came to an agreement – six. Because there isn’t something like one or three or five in her life.”*

Rina blamed herself for having allowed Wilna to start using sweeteners even though she was introduced to them by her friends. She continuously blamed herself and took responsibility for not realizing earlier that Wilna had been stealing food.

*“If I then knew what I know now, I wouldn’t have done it. I would never have done it. I would not have started with sweeteners because it was one of the ... [sigh]. We have terrible terrible, fights about that.”*

*“Yah! And but then I also I didn’t, and I wish I was more aware of that then.”*

Rina had always known that she had to restrict food access but she felt uncomfortable with it.

*“I should have then started to lock everything but I felt sort of ‘I don’t want to hurt her’.... It’s stupid. It’s stupid one mustn’t feel like that.”*

Rina eventually had to take a decision to restrict access to food. Control of food-related behaviours is complex, but centres on strategies to limit access to food such as consistent limit setting and close supervision at all ages including locking of cabinets and the refrigerator (IPWSO, 2013).

Rina further highlighted that when she finally decided to restrict food access by locking, she did not lock the deep freezer because she thought Wilna would not steal from a deep freezer. This was until she found out that Wilna was also stealing food from the deep freezer, and eating frozen and uncooked food. This agrees with the views of Gourash et al. (2006) who stated that food-seeking behaviour may include foraging for food in and out of the home, and also consuming spoiled, raw, frozen, or otherwise inedible foods.

*“So, while we are here, the storeroom is locked, I lock the pantry and also the fridge, because I thought the freezer, the freezer is safe ... The day I wanted to use that, she ate the pastry and I don’t know how she did it ... then the frozen chicken was still there ... and every time, then I think to myself ‘I don’t learn!’, because the other day ... it was beef and it was kind of a stew and I um took it out to defrost in the uh, uh, uh fridge, and there was gravy with it. Argh! I don’t know where I was and when I came back she ate the gravy part, and then I don’t say anything.”*

When Wilna steals food, Rina does not reprimand her, as she feels that it is her fault because she shouldn’t have left the food item unattended, or she should have

known. She takes full responsibility for Wilna's behaviour as she feels it is her responsibility to micro-manage her.

*"Because it's my mistake, I should have ... it is the parent and the carer's responsibility, cause they can't help it ... you have to be on the alert the whole time ... uh."*

Gourash and Foster (n.d.) emphasize that if we give a person an opportunity to steal food and they do steal food, then the plan is wrong, not the person. Hence there is no need for a consequence for food-stealing behaviour, unless the individual knows that you know about the food stealing because they were caught in the act.

Otherwise, it is advisable to act as though you have noticed nothing. Thus, the individual is not confronted with the evidence, and there is no consequence. But, behind the scenes, the family or the treatment team must review the procedure and make adjustments so that the opportunity does not occur again. Rina appears to have been applying this suggestion, and it appears to work for them. On the other hand, I believe it puts much pressure and responsibility on the primary caregiver.

Wilna does not have satiety problems. She is able to tell that she is full and can eat what is left over the next day. In addition to the lack of satiety problems, the fact that Wilna did not have food-related problems until the age of 18 was surprising and unexpected.

*"I think that was a kind of an uh, uh, uhm argh! She was fine till at certain stages and then that and I know it's different; they are not all the same. Some really have those craves, cravings from, uhm, and hunger feeling."*

There is much literature about satiety problems and food-related problems from the age of two in individuals with PWS. According to King (2008) two separate and distinct eating disorders are noted: initial feeding difficulties and failure to thrive, and then later overeating. In the second phase, the disorder continues with hyperphagia (excessive appetite for food), which seemingly begins in late childhood. The hyperphagia is hypothalamic in origin, resulting in a lack of sense of satiety. However, it was different for Wilna, and even the food stealing behaviour started later on.

Honey (2010) added that the changing eating pattern, which becomes evident as early as two years of age, causes major concerns, and then the over-eating and severe obesity become a central feature of the syndrome. The reason would appear to be a failure of the satiety response following food intake. The vast majority of individuals with PWS eat continuously and show no slowing down in their eating behaviour when faced with food. Once again, this was not the case with Wilna, as stated above, as she was able to get full and was even able to leave food for the next day.

#### **4.4.1.10 Temper tantrums**

Rina reported that when Wilna is stressed, it seems hard for her to contain herself and she has an outburst. It appears that she needs an outburst in order to express herself because an hour after an outburst she feels better. It was difficult for her in the beginning as she used to blame herself, but she eventually realized that the outbursts were not directed at her most of the time, and she has learned how to handle the outbursts.

*“That was one thing of Wilna. She, I think uhm, when she has emotions in her and stress in her she actually needs uhm ... [sigh], an outburst to sort of get rid of ... Then after an hour of outburst it is as if she feels better. Yah! And it’s hard for her to keep it inside ... as if it has to get out in a bad way.”*

This is in line with the results of the study done by Clarke et al. (1996) that suggested that temper tantrums, self-injury and impulsiveness are maintained as characteristic behaviours in PWS adult life.

#### **4.4.1.11 Weight control**

Rina declared that the only challenge they experience is around weight control. She would like Wilna to lose at least two kilograms. In order to achieve this, she takes her for walks and they weigh her every Sunday.

*“We have our challenges and uhm, uhm, what we do, we get on a scale every Sunday and uhm, I think, I want Wilna to lose a bit, but it’s not that easy ... and that’s why we go for our walks often.”*



On the other hand, Rina described Wilna as educated and disciplined when it comes to making certain food choices. She acknowledged that when Wilna has a choice between fried chips and salad she always opts for a salad.

*“You know she really likes her vegetables, so, um, I knew the weight she gained was in that time I wasn’t aware ... and I must compliment Wilna, she will never when there is a choice between chips and sandwich, or salad with a sandwich, she always chooses the salad.”*

Rina wasn’t sure if Wilna continues making the healthy food choices when she is not around and she is out with other people.

#### **4.4.1.12 Siblings**

Rina had ambivalent feelings about the fact that she does not have other children and feels there is no balance. On the one hand, she was grateful that she only had Wilna to worry about and that she did not have other children.

*“You know I have only one child, I think that’s the ideal, but on the other hand there is no balance. I think when you have more children in the house there is kind of a balance. You don’t concentrate the whole time and we are very much in a routine.”*

#### **4.4.1.13 Occupational life**

Rina further mentioned that after Wilna finished school she felt that she was able to work, but she did not want to go around asking people to hire her daughter. She then got a job coach, which was an occupational therapist who assisted people with finding a job. Drago (2006) stated that individuals with PWS can get involved in supported employment which is a community based job placement in an integrated work force. Supported employment is supervised by a job coach, who provides onsite job training and acts as a liaison with the employer.

*“You know, argh! When Wilna finished school, I, I felt, she is able to work and uhm, it was a bit of a problem because I feel it’s not my duty to ask people, ‘please don’t you have work or something for my daughter?’, so we got hold of uh, uh, a job coach.... We got the job coach and she was the person between the business and me.”*

Wilna was then employed twice in her life first at the library sorting books for a period of three years, and then at the post office. Some level of functionality was observed in the work place. Her job at the library ended because they terminated her contract because they experienced problems with her as she struggled to take orders from others and wanted to do things her own way.

*“Yah! yah! And sorting things out and so on. Uh, ok, so Wilna she worked there for about three years, but it was a lot of frustration ... she argh! Sometimes she wants to do things her way.”*

The environment at the post office was also stressful for her. She used to have terrible outbursts, when her boss who she was comfortable taking instructions from, was out of the office. Subsequently, she had to leave the job. This pattern has been observed in the family system as well, in that she can take orders and discipline from Rina but not from her father. According to Drago (2006) and Kazemi and Hodapp (2006), staff working with individuals with PWS get frustrated and have a low tolerance for various Prader-Willi behaviours. Behaviours characteristic of the syndrome, in fact frequently lead to job terminations.

*“As I told somebody this morning, in our house it’s the same. She does take discipline from me, but not from her dad, and this is something I’ve read in the literature as well. It was the same at the library, it was the same.... You know and, uhm, and she had temper tantrums there, shouting at people and – you know, and I still feel that I would like her to, to work, but definitely not in the open market.”*

#### **4.4.1.14 Independency**

It was evident that Wilna has some form of independence or was allowed some space to be independent in her life. Her occupational life, as stated above, was one such example and she also has her own bank card and can manage her own finances. She is also allowed to be on her own in a shopping mall. Even at church she attends a different service to her parents. I thought it was important that Wilna also embraces her individuality, as she tends to have such an enmeshed relationship with her mother.

*“Wilna has a debit card ... and, uhm, I allow her to be on her own in Brooklyn.... And then our church is the one here at Loftus. They have a*

*service for the families and Wilna goes to that service, and we're in the church, more the traditional service, but there, and I actually think that's so nice she knows people there, and families there, and people with babies that I don't know ... so that's part of her life and I think that is very special ... and I'm not part of that."*

#### **4.4.1.15 Hobbies**

Rina said that Wilna enjoys doing puzzles (see Figure 4.1, below), reading magazines and going to the baby department to look at baby clothes. She also enjoys colouring but she can only focus on one activity at a time. This has been observed of people with PWS, that they tend to withdraw into solitary activities for example, doing word puzzles and jigsaw puzzles, rather than activities with their peers (Whittington & Holland, 2010). Wilna also enjoys going to the dance that is arranged by the Down Syndrome Association, once a month. Rina said that Wilna is:

*"Busy building puzzles, she must show you."*

*"She likes reading the Beeld, she buys the Huisgenoot every week, and then, there is a magazine Baba en Kleuter, and you do get it in English as well as Baby and something."*

*"And ah, and then the Down Syndrome Association they have a sokkie, a dance once a month."*

#### **4.4.1.16 Fascination with babies**

Rina mentioned that Wilna at some point was curious about marriage and babies. However, they discussed the issue and Wilna realized that taking care of a baby is a lot of work. At present she appears to be fascinated with baby things, clothes, she reads baby magazines and likes asking her parents questions about their babies.

*"She likes going to Clicks and Woolworths to go to the baby department looking at all the baby clothes and so on ... and to go to a mother and talk to the mother about the baby and ask 'do you breastfeed?'"*

Rina seems to think that the fact that Wilna is reading baby magazines, in a way fulfils her biological need to have a child:

*"She likes reading. In a way that, uhm, I think in a way fulfils that feeling."*

#### **4.4.1.17 Social life**

Rina was worried that Wilna does not have friends. Her friends end up being Wilna's friends, and consequently she experiences their relationship as an enmeshed one. Furthermore, the dance that Wilna attends is part of her social life as she meets with her peers who also have genetic disorders.

*"That's a problem, and Wilna doesn't have friends but my friends, uhm are also her friends, I have one friend Bertha, she is an educational psychologist, she understands her and they really have a close, uhm, friendship."*

#### **4.4.1.18 Social support**

When I asked Rina about her support system, she struggled to answer the question, had a long pause, and actually seemed puzzled. She alluded to the fact that she lacks adequate support systems in her environment, and instead she is the one supporting other people:

*"Uhm this friend I mentioned who is also very dear to Wilna, but you know, uh huh, Molelekeng, my experience is [pause] I am actually, am supporting other people."*

Rina finds her strength in God, gets support from the people at church and from her one friend she considers her confidant. She further mentioned that her four siblings and their wives were also very supportive, and they check in on her from time to time and they visit each other:

*"Yah! That's one thing I must say, you know emm huh! I believe in God ... and I really, a lot of my strength ... my patience, and my strength, uhm, that's how I feel.... It's really something for me to go to church on a Sunday."*

#### **4.4.1.19 Marital support**

She remarked that her husband is supportive, that he is in the background because he is not a talkative person, and that he is not emotionally expressive but he demonstrates care for her.

*"And very seldom really shows his ... emotions, but he is there in the background.... You know, helping with other little things and so on, but not really, uhm, he can't really handle Wilna, uhm, he gets very, very upset when [pause], she, uhm, gets a tantrum and so on, because he feels she is hurting"*

*me when she is shouting.... He feels sorry for me, because he feels she is hurting me and he doesn't want me to get hurt."*

Rina spoke in a frustrated and disappointed manner as she continued to talk about her husband's support. She was disenchanted by the fact that her husband still does not understand their daughter, and has not found a way to handle her tantrums.

*"And then sometimes I feel, we have been in this situation for 33 years! And sometimes I feel he, he doesn't really understand her.... You know then sometimes I feel yhu! Argh! After so many years, the example I [pause] put to him."*

Throughout the interview with Rina, I found myself looking for her husband in her narrative, and wondering about his involvement. It initially came across as though he did not exist and then he was somewhere in the background. She sounded like she was in this alone, she was overwhelmed and lacked adequate support from him.

#### **4.4.1.20 Wilna's relationship with her father**

Although Wilna's father cannot handle her temper tantrums, they still have activities which they do together such as watching golf, motor racing and rugby. Rina does not like to watch sport so much, and maybe this is one situation where father and daughter can get closer to each other.

*"They watch rugby together."*

#### **4.4.1.21 Government support**

Rina mentioned that Wilna receives a disability grant.

#### **4.4.1.22 Need for structure and consistency**

Rina indicated that Wilna is very good with time and needs consistency in her routine. If she expects an activity at a particular time, and it does not happen, she gets upset. If there is a reason why the activity is not going to happen you have to inform her and reschedule it with her.

*"Say for instance, and she knew then on a Wednesday after say after break, I have speech therapy, on a Thursday then its physio and so then they didn't*

*come and fetch her.... Then say tomorrow they will come and says 'listen it's time for physio', and she refused. She will say it's not my time, you should have come and fetched me.... And they learned it's with Wilna you have to go and say 'listen Wilna, uhm, the physio has a special evaluation to do, she is very sorry but she can't take you now. Is it ok if we come and get you tomorrow morning?' Then it's fine."*

Wilna struggles to cope with sudden change, and therefore Rina spends time preparing activities in advance in order to avoid such changes. She constantly has to negotiate and discuss with Wilna if there are any changes that will be made. According to Whitman (1995) good management means always anticipating tomorrow, next week, new situations, changed plans, and the like. Clearly delineated authority figures and anticipated rewards and consequences provide the structure for behaviour, with clear limits without ambiguity. This is what Wilna requires, because she functions better within a structure. This once again requires the primary caregiver to work hard.

*"Change, sudden change is still a problem, you know ... yah, and discussing, 'listen is it ok if we go to Brooklyn, say Thursday morning to do this and that' or is it ok, say we've decided on Thursday and suddenly 'Wilna we can't go on Thursday, now what do you think? I think we should go Saturday' or you know and ask her ... and my whole life is, uhm, preparing."*

#### **4.4.1.23 Future plans**

Rina did not have future plans for Wilna, but she would like her to have a part-time job but not in an open market as she struggles to control her tantrums, and co-workers and customers will not be able to tolerate her. This was discussed previously in her occupational history.

*"I would like Wilna to, to say for instance to be involved, say, for two mornings or three mornings somewhere."*

#### **4.4.1.24 Psychological support**

Rina felt that they did not have a need for psychological support, as they had established an effective routine and everything was under control. However, it still is not always easy to handle the situation with Wilna:

*“I think, uhm, we are ok but I don’t think one can ever be [truly] ok.”*

Rina’s perception of psychological support contradicts Moss (2009) and Whittington and Holland (2010) who found that heavy demands are made on parents and caregivers of individuals with PWS which usually leads to negative experiences and feelings. Subsequently, families of people with PWS are subject to stress, and 70% of mothers have high levels of stress which need psychological counselling.

#### **4.4.1.25 Awareness**

In our discussion, it was evident that most people do not know about PWS. These people include lay persons as well as health professionals. Rina, together with other members of the PWSSA, are trying to invest in awareness. Rina mentioned that she recently had a presentation at Selwyn Segal Centre, with the aim to raise awareness to the audience, amongst them were health care professionals. She raised awareness by sharing her lived experiences.

*“It was attended by about 36 to 40 persons, many of them from Selwyn Segal, and then from outside as well ... and there was, uhh also a black lady, a psychologist, uhh, uhh a psychiatrist.”*

Despite the lack of knowledge and awareness, there also seems to be a gradual and growing interest from people who want to know more about PWS:

*“And they got this uhhm, uhhm, uhh ... assignment to do something about genetics and she remembered Prader-Willi Syndrome.... So, they interviewed me and they interviewed Wilna, more or less [a] very, very easy questionnaire.”*

She also mentioned that as the PWSSA, they make use of Facebook to raise awareness.

*“And Verda – she put stuff on the Facebook of the Association.”*

The challenge to reach communities, including rural areas was a reality. It was apparent that the Association needs to invest in awareness campaigns. In our overall

discussion it was highlighted that there is a huge need to ensure most people are informed and educated about PWS.

In order to achieve one of the aims of the study – to increase knowledge among other healthcare professionals during the International Prader-Willi Syndrome month, I arranged with the Sefako Makgatho University radio station, for Rina to give a talk. The radio station has an audience of students who are *inter alia* studying medicine, physiotherapy, occupational therapy, nursing, the sciences and speech therapy which I would regard as one of the stepping stones towards raising awareness.

#### **4.4.1.26 Knowledge about other families**

During the interview, Rina revealed information about other families who are members of the PWSSA, and it seemed that she knew about their challenges. For instance, she told me about Irene, whose husband passed away (Irene is discussed in Chapter 7). She also told me about Anna and Dikeledi who do not have access to PWS information due to a lack of computers and internet resources (Anna and Dikeledi are discussed in Chapters 6 and 8). Rina also informed me about a family in Johannesburg who were struggling with controlling their son's weight and behaviour. She was hoping that I could come up with an intervention to assist this family but the PWS son passed away before I could meet with them. This highlighted to me that Rina had an invested interest in other families and was always willing to assist them with their struggles. This also suggested to me that the PWSSA members were a close group of people, who knew each other and were, in an undocumented way, providing support to one another.

#### **4.4.2 Themes identified from interviews with Wilna**

The interviews with Wilna were difficult to conduct, as she struggled to express herself clearly due to speech difficulties, and because of the illogical, non-systematic flow of her content and the fact that she sometimes moved from one topic to the next and jumped logical levels. Although I felt out of my depth, I tried to remain calm and collected. The following themes were identified from our discussions:



#### **4.4.2.1 Diagnosis**

Wilna grew up knowing that she had PWS, but finds that other people diagnosed with PWS struggle to understand the syndrome.

*“Yes, yes. But for most of our children, it’s very difficult to understand the syndrome.”*

#### **4.4.2.2 Food-related problems**

She stated that some of the challenges she experiences are related to food, but did not elaborate on this, and just briefly said:

*“About the eating.”*

#### **4.4.2.3 Weight control**

Wilna mentioned that she is informed about healthy eating and always tries to make the right food choices.

*“When I go with people out for some breakfast, when I know I like my muesli, my yoghurt and my fruit salad, then I am ok with that type of food. I never eat the bacon, eggs and all that fatty foods.”*

Wilna is expected to lose weight, but she asserted that it is not easy for her to lose weight and her mother expects her to lose weight quickly.

*“You know, she wants me to lose quicker, but I take it how I am ... but it’s not that much easy. It’s not that easy ... as my mom thinks it is.”*

#### **4.4.2.4 Need for structure and consistency**

Wilna stated that she finds it difficult when she is not prepared and does not know what to expect. She gets frustrated when there is a lack of structure or routine.

*“And places we go, but we don’t know what to expect.... No structure, yes yes.”*

According to Cassidy and Driscoll (2009) characteristic behavioural patterns begin in early childhood in 70-90% of affected individuals and are typified by temper tantrums, stubbornness, controlling and manipulative behaviour, compulsive-like

behaviours, and difficulty with a change in routine. This has been reported to be the case with Wilna.

#### **4.4.2.5 Social life**

Wilna declared that she has a few friends and meets them when they go to the dance, and she also communicates with her one friend through short message services (known as “SMS”).

*“Yes, I have, but not that much.”*

#### **4.4.2.6 Schooling**

Wilna’s experiences of school were not pleasant, as she was teased and bullied by other children.

*“They bullied me, they used to take my bag and throw it on the floor – and do all the bad stuff.”*

#### **4.4.2.7 Occupational history**

Wilna stated that she enjoyed working, but that she experienced problems at Postnet because the person who accommodated her, and who understood and had accepted her, had left Postnet. When the replacement staff member arrived, she changed Wilna’s routine and nothing was the same. This sudden change was very frustrating for Wilna because she felt no one took her seriously, no one was listening to her, and she felt ignored. Butler et al. (2006) found that typical behaviour problems which include rigidity of personality, perseveration in conversation, tantrums, and non-compliance occasionally worsen during adulthood, which was the case with Wilna in her Postnet work environment.

*“Nice, but when I was at Postnet you know, the man who really knows me exactly the way, was away when the ladies coming in. She was moving my routine, nothing was the way it was.... It don’t [sic] work, everything was falling out of hands, my hands.... Yes! Yes! That is for me red lights. I told them when I say red lights, I told them, or something like that, and they don’t listen to me. They wasn’t [sic] listening to me if I say I can’t handle it. I was telling the truth.”*

If she were to return to the job market Wilna would prefer to work with people who understand that she has PWS and accept her; people who will listen to her when she tells them she feels she is going to have a tantrum, and who will understand and call her mother so that she can leave work because she is not in control of her emotions. She describes her tantrums as a “red light.”

*“Accept me, accept me, me with the syndrome, the way I am.... Yes, yes, but some places they don’t accept you how you are.... But that’s what I say from the beginning, my first place. Accept me how I am and what is [sic] difficult problems, ask me exactly where there can be a problem if I say there can be red lights coming ... don’t discuss some other things.”*  
*“Bad stuff if there is something bad – that we can discuss it. When I start exactly playing my card, throwing tantrums so that you can phone my mom, my mom quick, then she can come and fetch me. That isn’t too much to ask; then I can get quickly out of there.”*

This emphasizes what has been stated before in the literature that people working with PWS individuals struggle to understand some of their behaviour. Drago (2006) states that vocational placements are still difficult to achieve for individuals with PWS. Vocational providers have not had to live with the individual with PWS; many are slow to understand the seriousness of the appetite problems and emotional volatility associated with the condition. Employers are likely to perceive these behaviours as merely discipline problems rather than as a natural manifestation of PWS requiring workplace adaptation.

#### **4.4.2.8 Temper tantrums (outbursts)**

Wilna emphasized that she is unable to control her emotions, and that her mother understands her better than her father. Her father cannot manage her outbursts. She would like him to be more understanding and give her space when she has an outburst as doing this helps her to feel calmer.

*“Sometimes I can’t handle stuff, my emotions.... She becomes sometimes tough but not that many [sic].”*  
*“Bad sometimes, he can’t handle my tantrums he can’t handle it.... He must be more understanding and give me that space that I can say the bad words, that, so that, cross stuff can come out of my mouth ... so that I can feel better.”*

Wilna would also like other people to understand and accept her for who she is:

*“That’s what people will never accept or understand, that is the one thing, if people can start ... to accept that and take it like that.”*

#### **4.4.2.9 Hobbies**

Wilna mentioned that she enjoys horse riding, which she does every Friday, and perceives it to be part of her weekly exercises. The way she described what she does when she is on the horse sounded very therapeutic even though she considers some of the exercises to be difficult to do.

*“I also like horse riding ... left foot, yes, but the opposite is the more difficult.”*

She also enjoys doing puzzles. Figure 4.1 (below) depicts puzzles and handiwork done by Wilna:

*“I build my puzzle.”*





**Figure 4.1. Examples of Wilna's puzzles and handiwork**

As mentioned previously, people with PWS may withdraw into solitary activities – for example, doing word puzzles and jigsaw puzzles rather than engaging in activities with their peers (Whittington & Holland, 2010). This has been observed to be the case with Wilna to date.

After the interview, Wilna took me to her room to show me how many puzzles she has, and the ones she has completed. The puzzles contain one thousand pieces in each box. She shared her strategy for building a puzzle, where she sorts the pieces based on their shapes and that makes it easier for her to complete the puzzle. I was amazed, and found her to be very creative. This was in line with the findings of Whittington and Holland (2017) that individuals with PWS have exceptional skills with jigsaw puzzles. They use the shape of the pieces rather than the picture or colour as their main strategy, and this advantage disappears when pieces are cut by straight line rather than traditional jigsaw shapes.

In Wilna's bathroom she had different coloured soaps and bath foams lined up. Rina informed me that when Wilna buys items she buys in twos or threes; she never buys one item. For example, if she buys tooth paste, she will buy two, and if she buys soap she will buy three of different colours. Wilna prefers colourful clothing and does not like dull or dark colours like black and brown.

Young adults with PWS seem to be prone to a variety of compulsive behaviours and some develop obsessive-compulsive disorder. Some behaviours that are classified as obsessions are: distress when changing routine; repetitive questioning; symptoms like hoarding; need to tell or ask; excessive showering, toileting, and grooming; repeated organization; writing; collecting; and the need to finish one thing before moving on to the other (Cassidy & Driscoll 2009; Honey, 2010; McCandless & The Committee on Genetics, 2011).

This obsessive behaviour is evident with Wilna, in that she has been reported to only focus on one task at a time and needs to complete it before moving on to the next one. Buying more than one item, the organization observed in her bathroom with lined up different coloured soaps, and the struggle to adapt to changes in routine all suggest obsessions.

#### **4.4.2.10 Skin picking**

Wilna also struggles with skin picking, which often happens when she is bored for example while she is in bed or watching television, and she reports that she does not have control over it. At times when she becomes aware that she is skin picking, she stops, but sometimes she continues. She has been skin picking for a very long time. When the sore becomes extensive, she stops, they treat it, and then she moves to another area.

According to Gourash and Foster (2005) the skin picking behaviour of patients with PWS has a wide range of severity from patient to patient, and sometimes in the same patient over time. Some patients have occasional minor skin picking, while others maintain large and open wounds. It has been related to boredom and anxiety, but objective evidence for this is difficult to establish. This appears to be the case with Wilna, as she reported that she skin-picks when she is mostly in her room alone in the evenings, and also in the mornings before she wakes up and starts her routine.

#### **4.4.2.11 Future plans**

Wilna reported that she does not have any future plans.

*“I don’t know, I don’t have any future plans.”*

#### 4.4.2.12 Psychological support

Wilna does not have a need for psychological support.

*“Everything is fine.”*

### 4.5 Interventions

- ***Routine and structure***

When Wilna was around three years of age, Rina had already established a scheduled daily routine, which has been maintained to date. They also plan in advance for activities that fall outside the daily routine. For example, when they travel or go on vacation they adhere to the routine. When they go to unfamiliar places they also prepare Wilna in advance, in order to avoid uncertainty and to eliminate outbursts.

It has been evident that with routine and structure Wilna functions better, and she gets frustrated when there is no routine and structure. The structured routine appears to be working effectively for this family, even though it requires the primary caregiver to work harder at maintaining it. Sondergaard (n.d.) and Whitman (1995) stated that the quality of life for the individual with PWS depends on the ability of caregivers to provide an environment that is structured enough for the particular individual. Failure to maintain an appropriate structure is debilitating for all concerned, but, most importantly, debilitating for the individual with PWS. The need to maintain a high level of structure and control in the lives of individuals with PWS places additional demands on parents and other caregivers. Caring for PWS individuals requires substantial understanding and patience, which is what seems to be happening in the family in this case study.

Gourash and Foster (2005) confirmed the effectiveness of structure and routine and stated that the core symptoms of the PWS personality are managed through an environmental programme called “the basic plan” – that comprises of a daily schedule, food security, and mandatory exercise. This further validates the effectiveness of this intervention.

- ***Behaviour therapy***

Two weeks after the initial interview when I contacted Rina about the radio show, she informed me that she forgot to tell me more about Wilna's outbursts. She further mentioned that my visit had a positive impact on Wilna. It was then evident that the caregiver needed an intervention regarding the outbursts which Wilna tends to have. Another appointment was then arranged to get more information and to possibly come up with an intervention.

The session concerned was held at the family's residence. They welcomed me warmly and Wilna was very excited to see me again. Rina introduced me to her husband Charles, but he did not participate in the session. During my check-in with Wilna, she was talking about the weather, then about de-worming, and also about her attempts to lose weight. I struggled to understand Wilna and to get the point of her narrative as she was jumping through logical levels.

I approached this session from a behaviour therapy perspective. According to Corey (2009) behaviour therapy practitioners focus on observable behaviour, current determinants of behaviour, learning experiences that promote behaviour change, tailoring treatment strategies, assessment, and evaluation. The aim of this session was to define the problem, identify what needed to be changed, to assess the effectiveness or ineffectiveness of the current intervention and devise intervention strategies. This is supported by Grant and Evans (1994) who advised that when facing a behavioural problem, one of the first things is to identify what is to be changed. They further emphasized that it is important for the behaviours to be clearly defined.

The primary caregiver requested to first see me alone, so that she could tell me about Wilna's emotional outbursts while Wilna continued with her puzzle. She began to tell me about the recent outburst they experienced. As she was describing the incident, Wilna came rushing out of her room. She was clearly upset with her mother and requested that her mother not tell me about the incident, as she felt it was personal, private, and did not want it discussed further. She felt that her mother was gossiping about her.



I was not prepared for this situation, but at the same time I understood why she was responding in this manner. To calm her and to normalize the situation I invited Wilna to join us. She emphasized that the information the mother was sharing with me was sensitive, and she did not want to talk about it again because it upsets her again. She emphasized that she wanted to forget about the incident. At this stage, Wilna presented as disappointed and upset and I was worried about the impact this would have on our relationship. Clinical and research evidence suggests that a therapeutic relationship, even in the context of a behavioural orientation, can contribute significantly to the process of behaviour change (Corey, 2009).

I created the context for Wilna by explaining to her again the purpose of the research study, and reassured her of confidentiality. Wilna was then put at ease, and she mentioned that it was difficult for her to control her outbursts, and that sometimes the emotions are too overwhelming and nothing could ever help her. Wilna struggled to comprehend the concept of finding an intervention. This was displayed in her resistance and stubbornness. This also further indicated a concrete thinking processes, which may impact on the way she receives and implements suggestions. I then adopted a client-centred approach (this will be elaborated later on in this chapter) where I empathized with her which mobilized her to elaborate on the triggers of the outbursts.

According to Wilna, the following were possible triggers: When:

- someone does not do what she wants;
- she was working at Postnet the new owner did not have time for her, changed her routine, and they did not listen to her whereas her previous boss understood her, discussed issues with her, and respected her privacy;
- people do not listen to her (she emphasized this); and
- she doesn't know what to expect.

It became evident in my discussion with Wilna and Rina that she genuinely does not have control over her emotions. Rina supported this by mentioning that she would say at times *"that's not me, I don't want to be like that"*.

- ***Social reinforcers***

Rina highlighted that Wilna attempts to control her outbursts when she is in unfamiliar places or in public, but when she gets in the car or arrives home she will then have her outburst. Wilna however emphasized that she can control herself sometimes, but sometimes she struggles to do this. I then used a social reinforcer by praising her, in order to encourage the behaviour of self-control. Grant and Evans (1994) state that social reinforcers involve the actions of other people for example response-dependent praise, smiles, kisses, and applause which strengthen desired behaviours. Subsequently, this facilitated a collaborative process where, together, we discussed the intervention in order to reduce or eliminate the problematic outbursts.

Based on the discussion, it was evident that the following have been effective thus far, as they have reduced the frequency of the outbursts:

- The fact that Wilna has stopped working.
- Being home most of the time in a structured environment has contributed to Wilna's calmness as compared to the time when she was working.
- Having a set routine.
- Rina has discussions with Wilna before she makes plans, and also makes her part of the planning process.
- Preparing in advance when going on an outing.
- Having a daily diet plan, and even when they go to a shopping mall it helps to know in advance what she can have while they are there.
- When she becomes calm after an outburst, they discuss it.
- Their close relationship has proven to be beneficial, as it allows them to communicate effectively.

- ***Rule-governed behaviour***

According to Grant and Evans (1994) changing behaviour with instructions is an example of what behaviour analysts call rule-governed behaviour. In full form, rule-governed behaviour is the behaviour of "following the rule". Rule-governed behaviour is often planned, calculated, logical, and it is often said that the person whose behaviour is rule-governed knows the rules. Based on the rule-governed behaviour, the following were applied, when they were experiencing an outburst:

- Rina does not argue with Wilna during an outburst.
- Rina remains calm during the period.
- Rina gives Wilna space to calm down.
- Wilna finds a private space to have the outburst whenever she is able to, as she is sometimes unable to control her emotions.
- Encourage Wilna's efforts in terms of self-control.
- Rina remembers that the outburst is not directed at her.

Implementing the above proved to be effective in reducing the intensity and duration of the outbursts. I also realized that the outbursts are far apart, Wilna does not hurt herself during an outburst, and the outburst seems to serve a function for her which is to express herself and calm down. Therefore, it would be difficult to find something to replace it. Instead, as part of the intervention, we decided that during their communication following an outburst, and when Wilna had calmed down, they can use the platform to identify the cause and to find solutions, in order to prevent outbursts triggered by the same event.

After implementing the suggestion, this approach proved to be effective as they were able to circumvent similar incidents by devising solutions for similar future incidents. For example, Wilna had an outburst following an incident that occurred in a shop where she told the cashier to separate her items from her mother's items, but the cashier rang them up together. Following this incident, they discussed the cause which was the fact that the cashier did not listen to her, and she wanted to be in control of her own money. They then came up with a solution to prevent similar incidents in the future which was that Wilna would pay for her items first or she would go to a different till.

The functional and close relationship between Rina and Wilna further facilitated the effectiveness of this intervention, as it made it easier for them to communicate effectively and to find solutions to the identified problem. After communicating about the outburst, Wilna is able to take a meta-perspective, and acknowledges that it was not necessary to behave in the problematic manner, and will apologise.

According to White (1998) any seemingly minor combination of life stressors can contribute to loss of control as evidenced by tantrums and self-injury. Once control is lost, it typically takes a period of time before control is regained. This is often followed by feelings of sadness, remorse and guilt. In addition, the following is suggested: be vigilant to life stressors and avoid or prepare the person for the change; read early signs of control loss and provide support; once control is lost, provide a safe area and wait it out; afterwards, provide a “talk-out”; and avoid negative consequences.

I then identified the attempts and the ability to see circularity in behaviour as a desirable behaviour that needs to be maintained and increased. This was done through label praise, where I commended Wilna on her attempts. This was very encouraging for her. Label praise (or descriptive praise), is praise that specifies the behaviour or features of behaviour that are desirable (Grant & Evans, 1994).

In order to keep Wilna motivated, to maintain the desired behaviour, and also to build up her confidence, I therefore used Wilna as a tool and empowered her to give advice to other PWS diagnosed individuals, especially children and she made the following suggestions:

- She finds it helpful to have a discussion with a parent, where she can say what makes her unhappy and what upsets her.
- It becomes difficult for her when she attends a function and there is a variety of foods to choose from, and she wants to taste everything, but she cannot and then she has to decide. In this situation, the individual with PWS and the primary caregiver must discuss what they are allowed to eat, and what they are not allowed to have, and she emphasized that it is better if the caregiver helps her to decide.
- She further added and emphasized that she would like people to be more understanding and sympathetic towards people with disabilities.

- ***Client-centred therapy***

A client-centred approach was adopted with this case, as stated earlier. According to Corey (2009) the client-centred approach (founded by Carl Rogers), emphasizes three core conditions: empathy, unconditional positive regard, and congruence. They

are not formulated as skills to be acquired, but rather as personal attitudes or attributes experienced by the therapist, and they need to be communicated to the client in order for therapy to be successful. If therapists communicate these attitudes, the client will become less defensive and more open to themselves and their world and will behave in prosocial and constructive ways.

The key characteristic of empathy is understanding another person's subjective reality as they experience it at any given moment. This requires an orientation toward the client's "frame of reference". Accurate empathic understanding implies that the therapist will sense a client's feelings, as if they were his or her own without becoming lost in those particular feelings (Corey, 2009). Unconditional positive regard refers to the experiencing and offering of a consistently accepting, non-judgemental and valuing attitude towards a client. The caring is non-possessive and it is not contaminated by evaluation or judgement of the client's feelings, thoughts, and behaviour as good or bad. Lastly, congruence implies that the therapist is real, genuine, integrated, and authentic. The therapist openly expresses feelings, thoughts, reactions and attitudes which are present in the relationship with the client (Corey, 2009).

In summary, Wilna seems to lack the ability to control her emotions, which results in outbursts. The outbursts occur infrequently once a month or once every four to six weeks. As long as the environment is controlled, however, Wilna rarely has outbursts. The meaning and function of the outbursts was identified. Understanding the meaning and function actually put the primary caregiver at ease and Wilna felt understood, which was encouraging for her. It was also evident that it would be difficult, if not impossible, to completely eliminate the outbursts. They could only try and manage them.

One of the aims of the study was to determine the effectiveness of the interventions currently in place. It was evident that the structure and routine seemed to work effectively for this family, as it helped keep Wilna occupied and managed the frequency of outbursts. The suggested ways of managing the outbursts and the communication that followed were also effective in preventing future outbursts related to the same triggers, and facilitated Wilna's ability to take a meta-perspective.

## **4.6 Conclusion**

This case highlighted that the primary caregiver dedicated most of her adult life to caring for her daughter and to PWS-related activities. She also had a vested interest in the challenges experienced by other families who are also members of the PWSSA. PWS basically took over her life. The case demonstrated that caring for an individual with PWS puts a lot of strain and responsibility on the primary caregiver, and it becomes an overwhelming and time-consuming experience. The primary caregiver has to constantly manage situations and the environment. Ensuring consistency and having a structured routine seemed to be very effective in terms of managing the individual with PWS.

Despite the above considerations, throughout the literature the focus has been on the individual with PWS as if the primary caregiver does not exist or have their own life. This must have a significant impact on the primary caregiver in this case. It was evident that the primary caregiver lacked an adequate support system, but her beliefs and faith in God help her to cope with the challenges she is experiencing. The husband does not seem to participate in Wilna's day-to-day management; he assumes a limited role, and this lack of support is frustrating for Rina even though she attributed it to Wilna's tantrums. It was interesting that given her lived experiences, Rina had no need for psychological support.

It was intriguing that Wilna had no satiety problems which are predominant in the literature, although she occasionally stole food when she had access to it. Subsequently, the primary caregiver had to restrict food access which left Rina with guilt associated with locking away the food. Rina also blamed herself constantly when food was stolen, because she perceived it to be her responsibility.

Wilna is constantly facing the struggle of uncontrolled emotions and the need for acceptance in the work environment and in society. Due to her temper outbursts it is a challenge for her to gain and sustain employment which further renders her dependent on the primary caregiver. Rule-governed behaviour, routine and structure, social reinforcers, and a client-centred approach have seemingly been effective in managing outbursts.

## **CHAPTER 5**

### **TSHEPISO'S STORY: "SHE IS NOT ABLE TO READ"**

#### **5.1 Introduction**

This chapter presents a case study of the lived experiences of the primary caregivers Makhotso and Thabo. Tshepiso, their daughter, has been diagnosed with PWS. This case study demonstrates the significant impact of raising a child diagnosed with PWS, the pain of not being able to undertake some motherhood activities, and the effectiveness of behaviour therapy as implemented by this family. This case will also illustrate the differences between Tshepiso's presentation and what is being documented in the literature. The findings are presented according to the topics discussed in the interview and the themes that emerged. The statements are supported by italicised and indented quotes from the transcripts. The findings are integrated with the literature reviewed in Chapters one and two.

I conducted two lengthy sessions which took approximately two hours each with this family. The first session was conducted with the mother, Makhotso, and the second session was conducted with Tshepiso, Makhotso, and Thabo (the father). The first session focused on the interview questions and the second session again focused on the interview questions, and also assessed the effectiveness of the current intervention based on behaviour therapy and provided psycho-education on the cognitive functioning of an individual with PWS in order to manage and minimize the pressure placed on Tshepiso. A brief follow-up session was conducted. The first and second interviews were transcribed and translated. The interviews were conducted in both Southern Sotho and English.

#### **5.2 Family background**

This family comprises the father, the mother, and one child. They reside in a small, but beautiful home in Vosloorus – a large township in Ekurhuleni, Gauteng. The area is mostly populated by the black African emerging middle class. The family environment is conducive for living. The combined family income is R11000 a month. As the family does not own a car, they depend on public transport. They are members of the Anglican Church. The father attends church every week; however, the mother and child rarely go to church. The reason for this is that they prefer to

catch up on household chores they are unable to do during the week. Their home languages are Setswana and Southern Sotho. They are affiliated with PWSSA.

### **5.3 The context**

The first interview was conducted at a restaurant and the second was conducted in their home in Vosloorus. They warmly welcomed and hosted me in their home, and it was a humbling experience as they were excellent hosts and prepared lunch for me. The primary caregivers appeared to be warm, loving and supportive of Tshepiso.

Makhotso is a married, 40-year-old African female. She has matric and is currently working full-time as an administrative clerk. She was welcoming, friendly and warm. She spoke about her experiences in an emotive manner, especially when she spoke about the diagnosis and the struggles Tshepiso is experiencing at school. However, she usually spoke in a relieved manner when she discussed Tshepiso's behaviour, as they were not experiencing any behaviour problems with her.

Thabo is a married 43-year-old African male. He is currently working as an architect. He was welcoming and friendly. He spoke about his experiences, mostly in an emotionally contained manner. However, his frustrations were evident when he spoke about Tshepiso's lack of interest in reading.

Tshepiso is a 13-year-old African female. She is currently a scholar in grade five. She receives a disability grant. She presented as friendly, warm, and came across as shy. She appeared taller compared to other individuals with PWS. She had observable dysmorphic features, gait difficulties, and also speech difficulties. The interview with Tshepiso did not take long due to speech difficulties, which meant she did not elaborate much on some of her responses.

According to Lewis (2006) the speech and language skills of individuals with PWS are reported to be below expectation, based on intellectual levels. The difficulties include poor speech-sound development, which includes errors due to poor motor abilities associated with the production of speech sounds and errors in applying linguistic rules to combine sounds to form words; reduced oral motor skills; and also, language deficits. Language problems include deficits in vocabulary, grammar,



morphology, narrative abilities, and pragmatics. This explains why Tshepiso was struggling with narrating and formulating sentences. She mostly used non-verbal responses and gave one-word answers. She was observed to be struggling to pronounce some words. I had to listen attentively or asked her to repeat words, in order to hear and understand what she was saying.

## **5.4 Lived experiences**

The lived experiences of the primary caregivers and the individual with PWS, Tshepiso, were explored by conducting semi-structured interviews with them. From the transcribed interviews, certain themes were identified, which are discussed in the following sections.

### **5.4.1 Themes identified from interviews with Makhotso and Thabo**

In the interviews with Makhotso and Thabo, the following themes were identified:

#### **5.4.1.1 Pre- and post-delivery**

Makhotso was 27-years-old when she fell pregnant with Tshepiso. She was healthy throughout her pregnancy. Although she had normal foetal movements, she gave birth prematurely at eight months through a caesarean section, because the baby was in a breech position. The birth took place in Baragwanath Hospital in Gauteng Province and she was satisfied and impressed with the level of care she received.

According to Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) hypotonia is prenatal in onset, and usually manifests amongst other characteristics as decreased foetal movement. This was, however, not the case with Makhotso. These authors also mention that sometimes there is an increased need for assisted delivery or caesarean section which was the case with Makhotso. However, this appears to have been because of the foetal position rather than hypotonia.

*“It was alright, it was normal.... Yes, I felt the movement and everything was fine, it was alright, there was nothing that was odd ... so, [at the] last moment at around 8 months she had not turned, and it was a breech.... It was a caesar.”*

Tshepiso did not cry immediately after birth and was very floppy. However, she quickly gained muscle strength, as, within a week, they reported significant improvement. Based on this and the information above this suggests that the hypotonia with Tshepiso was milder. The doctors further explained to them that Tshepiso would not develop like any other child and that her milestones would be later than normal.

*“When she was born, I had that thought, I did not hear her cry, but I think they beat her and then I heard her cry.... They took her immediately but they did not tell me anything ... they had not told me, what is what, what was going on, but they had put her in the incubator, the doctors kept coming. When they touched her, she was like an elastic band, she was loose, loose, loose. I was wondering if she would ever get strong, [and] after some time they explained that she will be fine but everything will be slow, she won't be like any normal child ... so for sure after like five days, four days, after a week, it was only then I actually saw changes that she would then cry when she was hungry ... she started opening her eyes.”*

#### **5.4.1.2 Feeding problems**

Difficulty in sucking is one of the most common symptoms of new-borns with PWS. Because of this, Tshepiso was fed through a feeding cup. Assisted feeding through a feeding tube and/or special nipples with increased feeding times are necessary for a period of time – usually weeks to months. In the past, some parents have fed new-borns with a dropper or spoon to ensure adequate calorie intake, while many have chosen to introduce cup feeding early in order to circumvent the inadequate suck reflex as was the case with this family (Cassidy, 1988; Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

Infants with PWS do not spontaneously demand feedings, as they rarely wake up to feed. Therefore, a regular feeding schedule should be established and the infant's diet must be adjusted as needed in order to maintain appropriate weight gain (Cassidy & Driscoll, 2009). Subsequently, Makhotsa had to wake her up every two hours to feed Tshepiso, as the infant would not wake up on her own. The feeding problems seem to have persisted throughout her early childhood, as she had to be closely monitored when eating.

*“She was not able to suck. I used to feed her through a cup like this one [demonstrating]. I had to open her mouth to feed her. She was always sleeping, she would not wake up, she would not cry to be fed. You had to think that she is probably hungry.”*

*“When I was raising her, I did not experience many challenges. The only thing was that when she was eating, I had to monitor her. Because when she was eating it would be as though she wants to vomit ... I wasn’t supposed to give her a lot of food or give her hard food like solids.”*

#### **5.4.1.3 Diagnosis**

Tshepiso was not diagnosed immediately after birth, but the symptoms were suggestive of PWS. Tests were then done to confirm the diagnosis a few weeks later. Caldwell and Taylor (1988) gathered information from 12 families and found that at the time of birth a diagnosis of PWS was extremely rare, and this diagnosis typically only occurred when infants began to demonstrate the phenotypic characteristics; some were misdiagnosed or not diagnosed at all. Tshepiso was born 15 years after these findings, by which time there then seemed to have been an improvement in identifying the symptoms after birth and in terms of confirming the diagnosis. This improvement was confirmed by Whittington and Holland (2010), who reported that compared to 20 years previously, there was a generation of children who have been diagnosed with PWS, days or weeks after birth. Bar et al. (2017) found that the mean age at diagnosis was 18 days.

*“From birth, because she was born very floppy and the doctors were concerned why she was so floppy ... I gave birth at Bara, so they conducted tests and they found out that it was Prader-Willi Syndrome.”*

The diagnosis did not immediately have an impact on Makhotsi, as she underestimated the seriousness of the condition.

*“It was my first child, so I wasn’t scared. I thought that she is going to be alright. They told me she will be slow, so I took it she is going to be slow, but my mind was telling me that she will be ok.”*

The family have not told Tshepiso about her diagnosis. They were waiting for her to get older, as they felt that she may not understand at present.

*“She doesn’t know we haven’t told her.... She is on the injection, but she doesn’t know for what purpose ... I also don’t know what to tell her ... I don’t know; maybe I will explain to her when she is a bit more mature.”*

#### **5.4.1.4 Genetic counselling**

Makhotso mentioned that during genetic counselling they were informed that Tshepiso would be intellectually disabled, have excessive weight gain, excessive appetite, and also behavioural problems. They were expecting that they would observe and experience all these issues while taking care of Tshepiso, but this was not the case. This resulted in them being worried that there was something wrong with their child, as she did not present with all the symptoms and behaviour characteristics as predicted. Tshepiso is mostly presenting with significant cognitive impairment. Caldwell and Taylor (1988) mentioned that children with PWS were given a poor prognosis with high probability of having significant cognitive limitations.

#### **5.4.1.5 Milestone developments**

As Tshepiso was growing up, they realized something was wrong, as some of her milestones were not reached and some were delayed. This was supported by Cassidy and Driscoll (2009) who stated that there are delayed milestones, including gross motor and language delays, in individuals with PWS.

*“As she was developing, you could now tell that something is wrong, she took time to walk, she took time to crawl. She first crawled on one knee; that is when I realized this is how she is ... even my family members were surprised, because it was the first time they see a child like this one.”*

Her speech was delayed and she used sign language to communicate. Her speech has significantly improved; however, there are still words that she cannot pronounce.

*“She was not able to talk properly; she used sign language to communicate.”*

Tshepiso is now developing facial acne and they attribute this to teenage hormones, but at the same time they believe she will start menstruating late because that is the information they got from the doctors during genetic counselling. They were told that they have to wait for the breasts to develop first, and one of her breasts has been

developing, together with pubic hair. To prepare her, Makhotso has already begun teaching Tshepiso about menstruation and how to use pads, and they are also being taught about this at school.

#### **5.4.1.6 Treatment received**

Following the diagnosis and genetic counselling they were referred to a dietician in order to manage the weight problems, to a speech therapist due to the language delay, and also to an occupational therapist and physiotherapist due to delays in crawling and walking. They had continuous follow-ups at the hospital. This was in line with Cassidy and Driscoll's (2009) suggestion that children with PWS should receive early intervention (including physical, occupational, and speech therapies), and also individualized appropriate education. They were even advised to apply for financial assistance from government.

Contrary to Whitman's (2006) research findings that parents of children with PWS reported higher levels of parental and family problems, suffered greater pessimism, and yet got less support from professionals when compared with the parents of similarly aged children with cognitive deficits from other causes, this family received adequate support from healthcare professionals.

*"Then the doctor made us aware of the weight gain on her thighs and told us not to over-feed her. We then went to the dietician, physiotherapy, OT – all those things."*

From the age of seven, to date, Tshepiso has been receiving growth hormone therapy every month from Baragwanath Hospital. Makhotso injects her every day at six o'clock in the evening. Thabo has not gathered the strength to do it, and therefore when Makhotso is not there to inject Tshepiso they miss a dose.

McCandless et al. (2011) and Whittington and Holland (2010) state that clinical experience suggests that growth hormone treatment can be beneficial for an individual with PWS, as early as two to three months of age. With growth hormone supplementation, many affected children have normal growth trajectories and a final height compatible with parental height. Treatment intended to increase height needs

to begin before the normal age of puberty which was the case with Tshepiso. It has also been reported that GHT assists with weight control.

*“It helped a lot with her weight.”*

#### **5.4.1.7 Medical problems**

Tshepiso’s overall physical health is good, and they attribute this to growth hormone therapy.

*“Ever since she has been on growth hormones she no longer gets sick. Even flu does not affect her much – when she gets it is for two days and she recovers.”*

Tshepiso still consults with her doctor at Baragwanath Hospital, and she has regular quarterly check-ups. They screen for diabetes, hypertension and also do a spine test. Tshepiso is currently experiencing sinus problems, and they plan to consult with an ENT (Ear, Nose and Throat) specialist.

#### **5.4.1.8 Impact of PWS on Makhotso and Thabo**

Makhotso and Thabo were constantly in hospital, moving from one professional to the next. It was strenuous for them, and this contributed to them delaying having another child.

*“I am scared of this whole procedure, but now I have told myself that if it happens I am ready. But, at the time, I was scared, wondering what if I get another child like this one, you see.”*

Makhotso felt deprived of the experiences of motherhood – such as breastfeeding.

*“You know, my experience, there are things I experienced, I wished that my child could breastfeed and feel what breastfeeding is like those little things. But then again you wonder what if I get a child like this one or get one who is worse than this one, you see.”*

#### **5.4.1.9 Decision to have another child**

Makhotso and Thabo have now made a decision to try for another baby.

*“Now I am ready [laughter] if it happens it’s ok ... I don’t have a problem anymore ... I have even stopped contraceptives.”*

Tshepiso seems to be ready to have a sibling as she has indicated a few times to her mother that she must have another child.

*“When she sees a child on TV, she will show me and tell me I am left behind.”*

#### **5.4.1.10 Schooling**

Makhotso and Thabo are worried about Tshepiso’s performance at school. She had to repeat grade R, because the teacher felt that her performance was not satisfactory as she was unable to grasp concepts and was slow compared to other children. As a result, Tshepiso started grade one at the age of eight and was enrolled in a special school. So far, she is struggling to write sentences and to read. She is also two grades behind her peers. The literature confirms that mental retardation has been considered an integral part of PWS, since it was first described in 1958 (Whitman, 1995). Cognitive disability is evident when they start school. Regardless of the IQ score, most people with PWS have multiple, severe, learning disabilities and poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

*“She is doing well but she seems to be struggling in school. I don’t know if it’s the school but she has been in the same school since grade one.... But she is unable to ... if I ask her to write a sentence: ‘I am going to school’, I need to spell it out for her, [and] only then she will be able to write it. But if I just say ‘Tshepiso write: I am going to school’, she won’t be able to write it [on her own] ... I know that my neighbour’s child is exactly the same age as Tshepiso, but she is in high school.”*

Makhotso and Thabo spoke about Tshepiso’s school performance in a distressed manner. They are concerned about Tshepiso’s future performance. They worry that she may not be able to cope in higher grades and wonder if she will ever get employment in the future. They have now resorted to forcing her to learn how to read.

*“That is a serious problem, it worries me. I wonder how she will cope in high school?... It’s like I told you, she is scared to read, I observe her getting so frustrated.... They can’t reach high school, I doubt if they can*

*get to high school... The problem we will experience is that if we leave her, she will never learn and if she fails to understand she will fail at school."*

Both Thabo and Makhotsa are worried about the fact that Tshepiso does not like to read, and they perceive it as lack of interest and manipulation.

*"She doesn't want anything that has to do with reading. She would rather be on her computer ... she gets irritated and she does not seem to be interested ... if she had an interest she would probably be able to read."*

Tshepiso also seems to be struggling with poor short-term memory:

*"You teach her something now, and after two minutes she has forgotten."*

Even though Tshepiso cannot read and has poor short-term memory, she seems to be able to memorize.

*"She is able to memorise, but in a different way ... every Friday they write [a] class test, so they will give them words that they must study next week. She will be practising those words the whole week and you will be sure that she knows those words ... only to find that she has memorised those words."*

Tshepiso receives adequate support from the school. When she misses school due to her regular check-ups on days they are writing a test, she gets a chance to re-write the test concerned.

#### **5.4.1.11 Food-related problems**

Makhotsa reports that during early childhood Tshepiso used to steal a slice of bread when she woke up in the morning, but that behaviour has since stopped, especially when she is home with them. Tshepiso now tells her when she is hungry, and waits for her to prepare food for them. Even when Tshepiso used to steal food, she would only steal a small portion. She also used to accept food from other learners, but after being reprimanded she stopped doing this. Makhotsa told her:

*"If someone gives you their food, you must tell them to take it back home."*



Makhotso attributes this desired behaviour to their “strict” parenting style. However, when Tshepiso visits Makhotso’s family, she does sometimes get complaints that she stole a slice of bread, but her behaviour is not out of control.

*“Tshepiso’s behaviour – I don’t know, maybe it is because we are harsh, because she is able to listen to us, but when I am not around she misbehaves. But when I am around she doesn’t do certain things, but during school holidays I take her to my family ... they would call me and tell me she woke up in the morning and stole bread and they didn’t see her.... She used to do it before, but it changed as she grew up, she stopped. She no longer steals food.”*

They do not lock food away, but Makhotso marks the content level of items she could possibly eat, so that when Tshepiso comes back from school she does not eat such items.

*“Even these things that they wake up early in the morning and steal food from the refrigerator – like I said earlier, she used to do it, but she doesn’t do it anymore. She does attempt sometimes when she comes back from school, but I will tell her, for example, that ‘I have left a cold drink and I have marked it, [and] if I find that it is below this line I will know that you drank it – so do not take this cold drink and drink it’. She used to steal something, but she would not consume the whole thing. She would take a portion and that’s when you will notice that she ate it.”*

Makhotso also uses scare tactics to control the stealing behaviour:

*“She knows that Thabo will shout at her so I scare her that ‘if you steal again, I will tell Thabo.’”*

Tshepiso seems to be disciplined and has control around food, even when she is unsupervised.

*“When I come back from work I will ask her what she ate when she came back from school, and she will say a slice of bread, and when I look in the bread bin, indeed, I will find out that she only ate a slice of bread.”*

*“I give her four slices, water and juice, and she tells me that during the first break she eats two slices and she has the other two during the second break.”*

#### 5.4.1.12 Behaviour problems

Makhotso reported that Tshepiso used to present with disruptive behaviour. When she was still young, she would ask a lot of questions from people and could not sit still.

*“She used to be disruptive as a child, but currently she is quiet and I prepare her in advance when we are visiting people – ‘we will be visiting people and when we get there you must be quiet, sit still and when they offer you food, take what they give you and don’t ask for more. Don’t stand up and touch people’s cupboards, otherwise we will go back home’ .... I also keep an eye on her. I tell her ‘if you do this I will leave and I am taking you with me. You must not embarrass me’ – and she listens.”*

At present Tshepiso does not display any behavioural problems at home or at school that they are seriously concerned about; she is well-behaved.

*“She is well behaved. Even at school I would ask if she is not naughty, and the teacher will tell me they don’t have problems.”*

On the other hand, Thabo and Makhotso mentioned that Tshepiso tends to be manipulative and stubborn at times. They did not speak about this in an overwhelmed or distressed manner, which suggested that the behaviour was manageable and did not require intervention.

*“But she has this thing, when she wants something, she wants it, she is stubborn. When I say ‘leave that alone, don’t do that’, she will answer in a cheeky way ... she won’t do things at the time you require her to do them, she will tell you that she is tired.”*

When Tshepiso washes dishes she takes a long time, but she will do it properly and neatly.

*“For example, when you tell her to wash dishes she will take almost two to three hours washing dishes.”*

Overall, Tshepiso seems to be able to take instructions from her parents and behaves accordingly. When she is told not to do something or to act in a certain way, she listens and responds accordingly. She can distinguish good behaviour from bad behaviour. The parents worry at times that Tshepiso might change and they may then experience behavioural problems.

*“We worry though, if she is not going to change.”*

A structured environment is more effective. According to Whitman and Jackson (2006) the structure should include: 1) family and house rules regarding what is expected, acceptable and unacceptable behaviour, along with specified consequences; and 2) schedules that are invariant in time and task routines. Consistency between parents or caregivers is also critical, since inconsistency invites frustration and acting out. In addition, the structure must anticipate and provide carefully designed opportunities for the individual to make choices. From the above narrations of the parents/caregivers, it is evident that this family has implemented these guidelines and they have also been very effective.

#### **5.4.1.13 Physical challenges**

Tshepiso struggles to run and jump and does not participate in any activities that require these abilities. She seems to tire easily and lacks momentum. She plays netball at school but when there are tournaments she only gets to be a cheerleader. As reported by IPWSO (2013) weaknesses in muscle tone, strength and motor planning skills make it difficult to gain co-ordination and speed for PWS children to partake in normal childhood activities and competitive sports.

#### **5.4.1.14 Sleep problems**

Sleep problems are commonly observed and reported to be one of the characteristics of PWS individuals. Excessive daytime sleepiness, day naps after five years of age, sleep apnea (a type of sleep disorder characterized by pauses in breathing or instances of shallow or infrequent breathing during sleep), restless sleep, and excessive night-time sleep are characteristic (Cassidy & Driscoll, 2009; Honey, 2010). Similarly, Tshepiso had major sleep difficulties when she was an infant, but now she has good sleeping patterns. She sleeps throughout the night and does not sleep during the day.

#### **5.4.1.15 Weight control**

Makhotso tries to control Tshepiso's weight by giving her small meals, and she often gets satisfied and will wait until the next meal. She may have a fruit in between

meals. Makhotso also monitors the amount of sweets she eats, because Tshepiso is allowed to go to the tuck shop and buy sweets for herself. This is in line with suggestions made by IPWSO (2013) that it is imperative to begin good meal management and education at an early age. This includes sticking to a strict schedule for meals and snacks, and limiting portion sizes. They also suggest that caregivers should instil confidence that the next meal will be served on time, by scrupulously maintaining mealtime routines.

*“I just give her two slices of bread. I don’t know if she gets full or not because I am mostly worried about weight gain.... She always wants sweets and everything, but I have to constantly monitor her and tell her not to eat too many sweets.”*

Exercise is a very important factor in weight maintenance, and early establishment of a routine of regular daily physical activity, of at least 30 minutes, is strongly recommended (IPWSO, 2013). In accordance with this, Makhotso and Tshepiso also exercise together, but they took a break during the winter season.

*“We used to take a walk around the neighbourhood. We also used to go to the park and gym there, but because I have become lazy I don’t go there anymore. The only thing we do is walk, but since it’s been winter we have not been walking.”*

#### **5.4.1.16 Satiety**

Tshepiso does not appear to have satiety problems. When she has breakfast, she will eat again around lunch-time and again around dinner-time. She does not ask for food in between meals. She seems to be satisfied with the amount of food she eats. This is contrary to Honey’s (2010) statement that the vast majority of individuals with PWS eat continuously and show no slowing down in their eating behaviour when faced with food.

*“If I give her two fat cakes in the morning I will tell her that because she ate fat cakes in the morning, she cannot have anything during the day. I will only give you biscuits, and that’s all, and she will only eat at night.”*

#### **5.4.1.17 Independency**

Tshepiso seems to be quite independent. When she comes home from school she is alone until the parents get back in the evening. She is able to prepare herself a light meal, wash dishes, and can do her homework without constant supervision.

*“She gets there, unlocks the gate; there is an alarm, so I showed her that when she arrives she must unlock the gate, unlock the burglar, de-activate the alarm ... She does not compromise on that; she has to do her homework, she won’t forget.”*

Thabo shared his frustration that on the other hand, even though it is not often, Tshepiso tends to go to her friend’s home and stay there until late. Makhotsa highlighted that this behaviour has since improved, and her friend actually assists her with homework.

*“After school she goes next door; sometimes she goes there in her school uniform. She will play until late and only come back when the mother is back. By that time, it’s too late and she is tired for her to do her homework ... But she is better lately, she doesn’t do it that much.”*

Tshepiso is also able to go to the tuck shop unaccompanied. However, she can only buy one or two items not any more. If she has more items to buy she will forget, unless you write the list of items on a piece of paper. She can take care of her personal hygiene. Her adaptive skills are, overall, good. When she is left alone in the house and wants to go and play outside, she has the ability to ensure that the house is locked, the key is safe, and that it does not get lost.

#### **5.4.1.18 Hobbies**

Makhotsa stated that Tshepiso enjoys playing with the smart-phone, copying paragraphs from a magazine, and drawing. She spends much time copying paragraphs from magazines and marking herself afterwards. She spends hours doing this. These behaviours have been observed in PWS individuals with obsessive-compulsive behaviours (repeated organizing, writing, collecting, need to finish one thing before moving to the next) (Ho & Dimitropoulos, 2010; McCandless et al., 2011).

The family recently got Tshepiso a puppy, which she also enjoys playing with. People with PWS tend to withdraw into solitary activities rather than undertake activities with their peers (Whittington & Holland, 2010). The above behaviour proves this, as Tshepiso mostly spends time writing, drawing, playing with her smart phone and with the puppy rather than playing with her peers.

*“Yho! She loves the tablet, she also likes magazines she likes to copy the paragraphs. She will copy the first word until the last word and when she is done, she will mark herself. She will draw and all sorts of things.... She loves writing.”*

#### **5.4.1.19 Social life**

Makhotso mentioned that Tshepiso prefers to play with younger children and not with her peers – because she tends to control the younger ones. Another reason is that she cannot play the games that her peers play – because she is not able to jump and run and becomes self-conscious as a result.

*“She plays with younger children, but she does not play with her peers. She likes the younger ones as I think she is afraid of the fact that the older ones are more mature than her. Maybe they will tell her to do this and that and she won’t be able to, so at least she is able to control the younger ones ... She is not able to do those things. That is why sometimes she does not like to play with her peers, because they will expect her to perform.... She has self-doubt.”*

Makhotso mentioned that she often discourages Tshepiso from playing with younger children but she does not seem to listen to her. This behaviour corresponds with what is mentioned in the literature that because social cognition may also be impaired, most people with PWS have difficulties relating to their peer groups and often prefer to be with older or younger groups (Whittington & Holland, 2010).

#### **5.4.1.20 Social support**

They receive adequate support from Makhotso’s family. After they were informed of the diagnosis they were empathetic and compassionate but at times they tend to struggle with the idea that Tshepiso has a restricted diet.

*“Her grandmother would say – ‘leave the child to eat, these doctors are just a bore.’”*

However, from Thabo’s side of the family they do not receive support, because the family believes that Tshepiso will be fine and needs to be treated like any ‘normal’ child.

*“They were not involved much because they were in denial. His sister would say that it would never happen, I must take her to a normal school, she is fine, as though we making up the story, And I have to explain to her that the doctor told us and I can’t take her to a mainstream school when I can also see her condition.”*

The couple have also deprived themselves of support from their friends, because they have not told them that Tshepiso has PWS.

*“I wouldn’t say from friends, because our friends do not know that she has Prader-Willi Syndrome ... I don’t talk about it, when I am with them I just tell them Tshepiso is fine, and that’s where it ends ... I don’t discuss a lot of things ... it stays amongst us.”*

#### **5.4.1.21 Marital support**

Thabo and Makhotso are supportive of each other. Ever since Tshepiso was born they have been taking her for her consultations together. The diagnosis and raising of a PWS child has not had a negative effect on their marriage. They had to accept that their daughter had PWS. Makhotso further remarked that her husband Thabo fails to understand Tshepiso at times, as he sometimes tends to believe she is acting out.

*“He is very good, but he has this stubbornness in him. We all know how Tshepiso gets, but on the other hand he expects Tshepiso to listen to him and do what he tells her to do ... he treats her like a normal person.”*

Caldwell and Taylor (1988) indicated that parents had different views on how the birth of the child impacted on their marriage. Some parents indicated it had no impact, while others mentioned that it strengthened their relationship as a result of mutual support as was the case with this family.

#### **5.4.1.22 Skin picking**

Tshepiso does not have serious skin-picking problems. Thabo and Makhotso mentioned that she has a tendency to play with a sore, but when she is told to stop she does so and never touches the area again.

*“Tshepiso does not have that problem. If she has a sore, I treat it and tell her not to touch it until it heals, and she does not touch it [again].”*

According to Gourash and Foster (2005) the skin-picking behaviour of individuals with PWS has a wide range of severity from patient to patient, and sometimes in the same patient over time. Some patients have occasional minor skin picking, as seems to be the case with Tshepiso, while others maintain large and open wounds.

According to Whittington and Holland (2010) no specific intervention has been uniformly effective. The behaviour is often extinguished if healing of the wound is achieved. Behavioural interventions have been effective in some cases. As stated above, when Tshepiso is told to stop, she does so, and this is therefore working for this family.

#### **5.4.1.23 Perseveration**

Tshepiso tends to ask a lot of questions and to talk about the same thing over and over again. However, when told to stop she does so:

*“She will ask you a lot of questions, over and over. She will also tell you a lot of stories without stopping. They currently have a school project. They have to collect 20 bottle tops; she now has 16, she needs 4, she was going on and on about it. I told her when you come she must not talk about that – she must sit and listen to you.”*

Typical behaviour problems observed in adolescence include rigidity of personality, perseveration in conversation, tantrums, and non-compliance which occasionally worsen during adulthood (Butler et al., 2006). In agreement with this, Tshepiso seems to only be presenting with perseveration in conversation.

#### **5.4.1.24 Government support**

Tshepiso is a recipient of a disability grant.



*“We applied for it, a while back. She got it when she was in grade one.”*

#### **5.4.1.25 Future plans**

Makhotso was overwhelmed by this question. She did not have future plans for Tshepiso, but she hopes she will become a sign language translator.

*“Eyi! Yha! nhe! I really don’t know ... yhu! I really don’t know. That’s why I sometimes feel like taking her to those schools where she can learn sign language, so that when she is looking for a job she can get work on TV, be one of those translators, something like that ... otherwise, I really don’t know what she will do.”*

Makhotso worries about Tshepiso’s life as she gets older. She wonders if she will one day get married and have children. She also shared her distress and worry about what would happen if she dies who would take care of Tshepiso?

*“I was wondering just the other day, when Tshepiso gets old, will she get married, what about the boyfriend, will he be able to manage her, won’t he notice that there is something cognitively wrong with her.... Will she have children, so you think about such things. You worry about what happens if God decides to take you before her, what is going to happen ... hey! You pray about it. It’s a problem. Yho! it’s a problem.”*

#### **5.4.1.26 Psychological support**

Makhotso felt that they needed support to manage Tshepiso’s academic difficulties.

*“If only she can read her school books; you would have helped us.”*

#### **5.4.1.27 Knowledge about other families**

During the interview Makhotso and Thabo referred to other families who are also members of the PWSSA. Makhotso compared her daughter to Stephanie (mentioned in chapter one and eight) who was my first PWS patient and who inspired me to do this study. She knew about Stephanie’s behavioural problems and delayed menstruation. She shared her empathy towards Anna (Stephanie’s mother), due to the possible challenges she faces due to behavioural problems. She felt that if Tshepiso was like Stephanie she would have struggled to cope. She also mentioned

Dikeledi and Kamo (see Chapter 6). It seemed that she knew about their challenges which she said were overeating and skin picking.

Makhotso also talked about Irene, whose husband had passed away (see Chapter seven). Makhotso and Thabo also informed me about a family in Johannesburg who was struggling with controlling their son's weight. All this information suggested to me that the PWSSA members were a close group of people, who knew each other, and who were in an undocumented way providing support to one another.

#### **5.4.2 Themes identified from the interview with Tshepiso**

The interview with Tshepiso was difficult to conduct as she struggled to express herself clearly. Due to speech difficulties, she did not elaborate on her answers. She typically gave one-word answers. The primary caregivers assisted by probing more when she gave her answers. She struggled to understand some questions, as she interpreted them concretely. The following themes were identified from our discussions:

##### **5.4.2.1 Diagnosis**

Tshepiso does not know that she has Prader-Willi Syndrome, because her parents had decided to not yet tell her about it.

##### **5.4.2.2 Sports**

Tshepiso reported that she plays sport at school. She does athletics and plays netball.

*"I play soccer ... I play netball."*

##### **5.4.2.3 Social life**

Tshepiso first told me that she does not have friends that stay in their neighbourhood, but, after the parents intervened, she told me she has one friend who she plays with during the day.

##### **5.4.2.4 Schooling**

Tshepiso stated that she attends Sparo Foundation School, where she started grade one. She agreed with the statements made by her parents that she does not like reading, and admitted that she cannot read.

*“Am not able to read.”*

She enjoys doing music, dance and art at school.

*“We dance and we sing arts [sic].”*

Contrary to her parents’ statement, Tshepiso claimed that when she misses school due to her regular medical check-ups, on the days they are writing a test, she fails the test.

*“Uhm, I fail.”*

#### **5.4.2.5 Sibling**

She stated that she would like to have a sibling that she would play with and take care of.

*“I will play with her.”*

#### **5.4.2.6 Treatment received**

Tshepiso mentioned that she gets injected by her mother every day, but she doesn’t know for what purpose. She commented that the father has never injected her, because he is scared to do so. She also reported that she takes flu medication occasionally, and visits the doctor every month.

#### **5.4.2.7 Hobbies**

Tshepiso mentioned that she enjoys watching television, especially movies, playing with her puppy, and writing stories that she copies from magazines.

*“I like watching TV ... playing with my puppy.”*

#### **5.4.2.8 Future plans**

Tshepiso reported that she would like to become a nurse when she grows up, because she likes injecting people.

### **5.5 Interventions**

- ***Psycho-education***

According to Lukens and McFarlane (2004) psycho-education is among the most effective of evidence-based practices that have emerged in both clinical trials and community settings. It is a professionally delivered treatment modality that integrates and synergizes psychotherapeutic and educational interventions. Psycho-education reflects a more holistic and competent-based approach stressing health, collaboration, coping, and empowerment.

A psycho-education model therefore views the role of the psychological practitioner not in terms of abnormality, diagnosis, prescription, therapy and cure but rather in terms of client dissatisfaction, goal setting, skills' teaching, satisfaction, or goal achievement (Authier, 1977).

According to Ahmed (2004) parents of children with intellectual impairment experienced depression, frustration, and were anxious about the future of their children. The frustration and anxiety about the future has also been this family's experience.

Family psycho-education includes teaching coping strategies and problem-solving skills to families, friends, and/or caregivers to help them deal more effectively with the individual. The rationale for psycho-education is that it reduces distress, confusion and anxiety in the family, which may in turn help the individual to improve their lifestyle (Ahmed, 2004).

Given the above considerations, the couple in this case study was psycho-educated on the cognitive functioning of a PWS individual, as they had concerns about their daughter's academic performance, were putting pressure on her to perform, and viewed her inability to perform as a stubborn and manipulative manoeuvre. The aim

of this intervention was to give the caregivers insight and to ease pressure on the individual with PWS.

A study on the parents of children with intellectual impairment, and who were assigned to a psycho-educational group intervention, showed significant improvement in parental attitude regarding child rearing and the management of the disability (Lukens & McFarlane, 2004). Following the psycho-education, the caregivers exhibited empathy towards Tshepiso, and understood that due to the cognitive impairment, she will always have limitations, and that all they can do is to try and support her and encourage her strengths instead of focusing on her weaknesses.

- ***Behaviour therapy***

I approached this session from a behaviour therapy perspective. According to Corey (2009) behaviour therapy practitioners focus on observable behaviour, current determinants of behaviour, learning experiences that promote behaviour change, tailoring treatment strategies, assessment, and evaluation. The aim of this session was to define the problem, identify what needs to be changed, assess the effectiveness or ineffectiveness of the current intervention, and devise intervention strategies. This is supported by Grant and Evans (1994) who advised that when facing a behavioural problem, one of the first things is to identify what is to be changed. They further emphasized that it is important for the behaviours to be clearly defined.

Tshepiso does not present with any behavioural problems. The reason for this is that the interventions currently being implemented have proven to be effective. Thabo and Makhotsa have been consistent in their approach in order to achieve and maintain desired behaviour from Tshepiso. It was evident throughout the interviews that Tshepiso's behaviour is managed effectively. The parents have implemented the following:

- **Rule-governed behaviour and anticipatory planning**
  - They are firm and consistent.
  - They give Tshepiso clear instructions to follow.

- They prepare her in advance when they have visitors or when they are going to visit someone, and warn her to behave.
- When Tshepiso does not behave, she knows that they will go back home.
- Tshepiso is told in advance about the consequences should she misbehave.

Some of the above interventions where instructions are used highlight the effectiveness of rule-governed behaviour. Rule-governed behaviour is often planned, calculated, logical, and it is often said that the person whose behaviour is rule-governed knows the rules. Changing behaviour with instructions is what behaviour analysts call rule-governed behaviour. In full form, rule-governed behaviour is the behaviour of following the rule (Grant and Evans, 1994).

The interventions the family are implementing agree with the behaviour management procedures suggested by Whitman (1995) particularly the anticipatory planning and consequences. In anticipatory planning, good management means always anticipating tomorrow, next week, new situations, changed plans, and the like; it also means pre-planned lunches, pre-stated rules, pre-determined and stated times to be spent in any one place, and with whom. Clearly delineated authority figures and anticipated rewards and consequences provide the structure for behaviour, with clear limits and without ambiguity.

Whitman (1995) emphasizes that the consequences of compliance or non-compliance need to be known ahead of time. In addition, consequences must be enforced without fail. Any change leads to the toppling of the whole structure. The family in this case study has demonstrated firmness and consistency – and hence this has been effective for them.

## **5.6 Conclusion**

It is evident that the primary caregivers struggled with Tshepiso in early childhood, but from late childhood they did not experience major problems especially regarding food-related problems like satiety and behaviour problems such as tantrums. They only experience minor incidents of manipulation and stubbornness that did not require intervention.

Tshepiso is tall and her weight is well managed compared to other adolescents with PWS which could be attributed to the growth hormone therapy. Tshepiso's presentation is also not typical of the individual with PWS, as described in much of the literature. Instead it seems to agree with Ho and Dimitropoulos (2010), that PWS is a spectrum disorder meaning not all symptoms occur in everyone affected, and the symptoms may range from mild to severe, as in Tshepiso.

It was clear that the cognitive impairment was a major concern for this family and there was a need for intervention. Psycho-education was offered and proved to be effective in reducing the frustration, worry, and anxiety experienced by the caregivers, and also reduced the pressure experienced by the individual with PWS.

It was again intriguing to discover that Tshepiso did not have satiety problems, which are so often mentioned in the literature. According to Goranson (2011) researchers strongly believe that the hypothalamus regulates appetite, metabolism, body temperature, and mood in people with PWS. It would be interesting to determine if there are individual differences in the hypothalamus of individuals with PWS, or if it changes over time.

Based on this case, behaviour therapy techniques where rule governed-behaviour was implemented and behaviour management techniques where anticipatory planning and consequences were implemented have proven to be effective. In addition, the possible link between parenting style, nurturing and improvement in desired behaviour also needs to be considered.

## **CHAPTER 6**

### **KAMO'S STORY: "IT'S MY WEIGHT"**

#### **6.1 Introduction**

This chapter will present a case study of the lived experiences of the primary caregiver, Dikeledi, and Kamo, her son, who has been diagnosed with PWS. This case study demonstrates the challenges they both face and the choices made by Dikeledi in parenting and managing Kamo's behaviour. Many of the choices and challenges could be attributed to a lack of education and information. The findings will be presented according to the topics discussed in the interview and the themes that emerged. The statements will be supported by quotes from the transcripts. The findings will also be integrated with the literature reviewed in Chapters one and two.

I conducted four sessions that took approximately one hour each with this family. The first session was conducted with the primary caregiver, Dikeledi, and the second session was conducted with Dikeledi and Kamo together. Both sessions focused on the interview questions and interventions. In the second session I inquired about the effectiveness of the intervention discussed in the first session. Two telephonic follow-up sessions were also conducted – that took approximately 30 minutes each. The third and fourth sessions focused on additional interventions and feedback on the interventions. The interventions were based on behaviour therapy and psycho-education regarding the cognitive functioning of an individual with PWS, in order to provide insight to the primary caregiver about the discipline techniques she could implement. I also referred them to paediatrics and psychiatry for pharmacological intervention. The first and second interviews were transcribed and translated. The interviews were conducted in both Southern Sotho and Northern Sotho.

#### **6.2 Family background**

This family comprises the mother and two children. Her husband and three of her children had passed away. The eldest son has his own home, where he lives with his family but he visits occasionally. Dikeledi and Kamo reside in a small RDP (Reconstruction and Development Programme) house that has one room which is divided by a curtain to make a bedroom, and then a living area with a kitchen. It has an outside toilet. The family lives in Lethlabile, a township in North West Province.



The area is mostly populated by black Africans of lower and working class. Their family income is R2800 a month. Prior to her receiving her pension in 2016, the family was surviving on Kamo's disability grant of R1500, and they were struggling to meet all their financial needs. They do not own a car and are dependent on public transport.

Dikeledi is a member of the 12<sup>th</sup> Apostolic Church and Kamo chose his own church and he is now a member of the Nthite church. Dikeledi attends church every week; however, Kamo rarely goes to church. The reason he gives for this, is that he wakes up late. Their home language is Setswana. They are affiliated with the PWSSA.

### **6.3 The context**

The first interview was conducted with Dikeledi in my office at the Dr. George Mukhari Academic Hospital on the day that she went to collect Kamo's growth hormone therapy medication. She was very accommodating, which I highly appreciated. The second interview was conducted with Kamo and Dikeledi at their home in Lethlabile. They fetched me at the mall; both seemed excited to see me. I was surprised that Kamo could recognize me, because I only met him briefly at the Annual General Meeting (PWSSA AGM). He was spontaneously telling me different stories.

At the mall I observed that workers and customers were greeting Kamo and starting conversations with him. It was evident that he was popular. I approached the sessions in a client-centred manner (this intervention is discussed in detail later on in the chapter). I was empathic, genuine and non-judgemental towards their circumstances, their socio-economic status and their home. Being client-centred put Dikeledi at ease and she welcomed me into her home despite the fact that she described herself as poor and not worthy of hosting me. Her response also resulted in me believing that she was experiencing felt stigma. According to Green, Davis, Karshmer, Marsh, and Straight, (2005) the stigma theory defined stigma as an adverse reaction to the perception of a negatively evaluated difference. One of the components of stigma is separation, which occurs when the reactions of others to these differences lead to a sense of "otherness", and this then leads to the experience of felt stigma.

Dikeledi is a widowed, 60-year-old African female. Her highest level of education is standard one (grade three). She did not continue with school because her mother did not want to educate girl children. She is currently a pensioner, and just recently started receiving an old-age grant. She struggled to recall information about significant background information. She gave contradicting information at times; however, when she was able to recall information whilst narrating, she managed to correct the contradictions, and at the end I could ascertain her overall experiences.

Kamo is a 13-year-old African male. As Kamo didn't know his age, he asked his mother. He is currently in a special school and has been in the same class ever since he started school at the age of 6. He receives a disability grant. Kamo presented as friendly, warm, spontaneous and talkative. These positive characteristics confirm IPWSO's (2013) statement which mentions that people with PWS have many positive characteristics, are known to be friendly, sociable, kind and caring, and many have a wonderful sense of humour.

Kamo has a short stature and is obese. He has observable dysmorphic features, gait difficulties, and speech difficulties. The literature suggests that children with PWS and who have been treated with growth hormone therapy (GHT) through childhood, can achieve normal adult height (Angulo, Butler, & Cataletto, 2015). There is also evidence from controlled clinical trials that GHT in children improves stature, muscle mass, facial appearance and bone density, head circumference, height, body mass index, body composition (with improvement of lean muscle mass and delay of fat tissue accumulation), body proportions, acquisition of gross motor skills, language acquisition, and cognitive scores (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Whittington & Holland, 2010). Contrary to these findings, and as stated above, there were no observable physical improvements with Kamo in relation to GHT treatment.

The interview with Kamo did not take long because I struggled to hear and understand him as his speech was unclear. He spoke in a hoarse and deep voice. He also had hyper-nasal speech. His tongue filled his mouth while talking, which made it difficult for him to form words. I could not understand what he was trying to say, and, at times, he used non-verbal cues to communicate. He did not elaborate much on his responses.

Kamo's presentation was in accordance with Lewis' (2006) research findings that voice characteristics reported for individuals with PWS include high pitch voice, harsh/hoarse voice quality, inadequate vocal intensity, and hyper nasality. Language problems also include deficits in vocabulary, grammar, morphology, narrative abilities, and pragmatics. During the interview when my attention was directed at the primary caregiver, Kamo sat quietly and did not interrupt.

## **6.4 Lived experiences**

The lived experiences of the primary caregiver and individual with PWS, were explored by conducting semi-structured interviews with them. From the transcribed interviews, certain themes were identified which are discussed in the following sections.

### **6.4.1 Themes identified from interviews with Dikeledi**

In the interviews with Dikeledi, the following themes became evident:

#### **6.4.1.1 Pre- and post-delivery**

Dikeledi reported that she was in her forties when she fell pregnant with Kamo. The pregnancy was unplanned and came as a surprise. She was healthy throughout her pregnancy. Dikeledi mentioned that she experienced foetal movement, but described her foetal movements as strange as there would sometimes be long periods with foetal stretching, and times when she would not experience any movement, and then she would worry.

*“Sometimes he would be so quiet that I would wonder if he was still alive.”*

This experience is in line with the reports by Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) who stated that in individuals with PWS, hypotonia is prenatal in onset, and usually manifests as decreased foetal movement.

Dikeledi's pregnancy was full-term and she had a normal vaginal delivery. This is contrary to the views of Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) who stated that in individuals with PWS hypotonia is

prenatal in onset, and usually manifests as abnormal foetal position at delivery, and sometimes there is an increased need for assisted delivery or a caesarean section.

*“Mhm, I was forty something.”*

*“It was strange, he would stretch for a while, if I am walking I had to stop.”*

*“Yes, I could feel that he was moving and playing.”*

In infancy, hypotonia is also characterised by decreased movement and lethargy, with decreased spontaneous arousal and weak crying (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010). This was particularly true in this case, as Kamo’s cry was reported to have been weak and soft after delivery. Dikeledi also noticed that the baby was floppy, and his neck muscles were very weak. The doctors examined Kamo and he was placed in an incubator but they did not explain the reason. After some time, the infant gained muscle strength, but Dikeledi could not recall when it happened. The birth took place in Ga-Rankuwa Hospital, now known as Dr. George Mukhari Academic Hospital in the Gauteng Province and she was satisfied and impressed with the level of care she received. They were admitted for two months.

*“The only problem was that he did not cry out loud as babies normally cry after delivery; it was as though his voice was stuck.”*

*“He was still a baby and was unable to lift his neck.”*

*“They did all the examinations, bathed him, and then put him in an incubator.”*

Dikeledi mentioned that she saw him just as any other baby but what was puzzling for her was that he did not have testes, but it seems the testes are now slowly developing.

*“He did not have testes. It looks like they have developed a little now.”*

Hypogonadism is another clinical feature of PWS, and occurs when the body’s sex glands produce little or no hormones. In individuals with PWS, hypogonadism has been observed in both sexes and manifests as genital hypoplasia (under-developed genitals) throughout life, incomplete pubertal development, and infertility in the vast majority. In males, the penis may be small, but most characteristic is a hypoplastic

(under-developed) scrotum that is small, poorly rugated, and poorly pigmented (Angulo et al., 2015; Cassidy & Driscoll, 2009; Cassidy et al., 2012; Hurren & Flack, 2016).

#### **6.4.1.2 Feeding problems**

Difficulty in sucking is one of the most common symptoms of new-borns with PWS. According to Dikeledi, Kamo could not suckle and they were forced to feed him through a spoon for two months until they were discharged from the hospital. According to Cassidy and Driscoll (2009), Ho and Dimitropoulos (2010) and Cassidy et al. (2012), part of the characteristics of hypotonia are poor reflexes, including poor sucking which leads to early feeding difficulties and poor weight gain. In the past, some parents have fed new-borns with a dropper or spoon to ensure adequate calorie intake (Cassidy, 1988) as it was done with Kamo. Dikeledi mentioned that after discharge she started her baby on soft porridge and infant formula, and he was eating well.

*“When I got back home I just stopped [laughter] because he wasn’t breastfeeding anyway. I was not going to continue force-feeding him. I bought him porridge, the smooth one ... Perlargon and porridge, he would eat.”*

#### **6.4.1.3 Diagnosis**

Dikeledi reported that Kamo was diagnosed while they were still in the hospital, after the doctors noticed that he was floppy. They tested him and established that he had Prader-Willi Syndrome. When they informed her, she accepted the diagnosis immediately; her husband and her family also accepted the diagnosis, and they were very supportive.

*“He was lethargic and floppy.”*  
*“When you play with a child who is a couple of days or a week old he can sort of stand, but Kamo couldn’t. They tested him and did everything and they found out that he has Prader-Willi Syndrome.”*  
*“What was I to do, I had to accept.”*

Caldwell and Taylor (1988) gathered information from 12 families and found that at the time of birth until infants began to demonstrate the phenotypic characteristics, a diagnosis of PWS was extremely rare; some were misdiagnosed or not diagnosed at

all. By the time Kamo was born, fifteen years after this publication, there seemed to be an improvement in terms of identifying the symptoms from birth, and also in confirming the diagnosis. This improvement was also reported by Whittington and Holland (2010) who reported that compared to 20 years previously, there was a generation of children who have been diagnosed with PWS within days or weeks after birth. Bar et al. (2017) found that the mean age at diagnosis was 18 days. Kamo's case suggests an improvement in the early diagnosis of PWS.

#### **6.4.1.4 Genetic counselling**

Dikeledi could not recall information given to her, as it was too much for her to remember all at once. She could only recall that they told her about obesity.

*"They just told me about obesity, but he was very tiny."*

*"I cannot recall, they were giving me a lot of information."*

#### **6.4.1.5 Milestone developments**

Kamo's developmental milestones were delayed; he took time to crawl, walk and talk. Cassidy and Driscoll (2009) state that there are delayed milestones, including gross motor and language delays in individuals with PWS. Early milestones are reached, on average, at double the normal age – for example, sitting at 12 months, walking at 24 months, and speaking words at 2 years.

*"When it comes to that, he used to attend physiotherapy, they used to give me exercises to train him, and they will tell me that now he is crawling, in the next month we want to see him stand. I was training him. When he stands this month, they will say the next month we want to see him walking. It took long. When he was walking he would fall, even talking, he is still struggling with that, it's just that I am used to him."*

#### **6.4.1.6 Treatment received**

Following the diagnosis and genetic counselling they were referred to a dietician in order to manage the weight problems, to a speech therapist because of the language delays, and to a physiotherapist due to delays in crawling and walking. They had continuous follow-ups at the hospital. This concurs with Cassidy and Driscoll's (2009) suggestion that children with PWS should receive early intervention

(including physical, occupational, and speech therapies) and also individualized, appropriate education.

Contrary to Whitman's (2006) findings that parents of children with PWS reported higher levels of parental and family problems, suffered greater pessimism, and yet received less support from professionals, when compared to parents of similarly aged children with cognitive deficits from other causes, the family in this case received adequate support from healthcare professionals.

Kamo has been going for quarterly check-ups at the Dr. George Mukhari Academic Hospital. Every month they provide Dikeledi with GHT and she injects Kamo three times a week; however, she could not recall when Kamo started GHT. They also screen for diabetes at every check-up.

#### **6.4.1.7 Medical problems**

Kamo used to have recurring influenza as a baby, but his overall physical health is good except for toothache and tooth decay. His milk teeth did not come out naturally, and they had to be extracted. Dikeledi mentioned that because Kamo is struggling so much with his teeth, she was even thinking of having all his teeth removed. He also recently had an ear infection.

*"As a baby, he had recurring flu. Am not sure what was the problem, I would take him to the doctor and he would get better."*

*"His teeth really bother him, he recently suffered from earache."*

*"I take him to the clinic when they are painful and they extract them."*

*"He has tooth decay. I was even thinking to have them all extracted and have false teeth."*

#### **6.4.1.8 Decision to have another child**

Dikeledi was advised by the doctors to sterilize immediately after giving birth to Kamo, and she was comfortable with this suggestion as she felt that she was old and did not want to have more children.

*"That's true. I did not want to have children anymore. I was old – what was I going to do with a baby [laughter]."*

#### **6.4.1.9 Schooling**

When Kamo started school, he was enrolled in a special school. Dikeledi mentioned that Kamo has been in the same class ever since enrolment and he cannot write. The teachers are struggling to teach him. Dikeledi showed me Kamo's report, and his marks were impressive as he got high marks for his subjects. However, when asked to write he simply scribbles and when asked to count he couldn't count past three.

These problems are confirmed in the literature; mental retardation has been considered an integral part of PWS since it was first described in 1958 (Whitman, 1995), and cognitive disability is evident when PWS children start school. Regardless of the IQ score, most people with PWS have multiple, severe learning disabilities and also poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

*"He went to a special school from the beginning."*

*"Kamo is not able to write."*

Dikeledi could not recall when Kamo started school and did not have adequate information regarding Kamo's education and performance at school. She did not seem to worry, as she was content to just send Kamo to school. I believe that this "laid-back" attitude could be because she underestimates the importance of education.

*"I don't know; as long as I take him to school."*

*"Ah, even if he says I am not going to school, I don't have a problem – I go back to sleep as well."*

#### **6.4.1.10 Food-related problems**

Dikeledi reports that during infancy and early childhood Kamo did not eat much, but he started gaining weight after the age of five when he started crèche because when he asked for more food there, they gave it to him. Dikeledi did not inform the teachers about Kamo's condition and possible weight problems. She perceived the weight gain as normal. In his current school, Dikeledi used to get complaints that Kamo ate other learners' food, and she asked the teachers to monitor him. However,



she no longer receives any complaints. Cassidy and Driscoll (2009) and IPWSO (2013) caution that lack of satiety should be acknowledged at school, work, and at home and all caretakers and supervisors need to understand this. Consistent limit setting and close supervision are necessary at all ages. Unfortunately, Dikeledi failed to comply with this despite having had genetic counselling.

*“Growing up he didn’t eat much at all, and he did not gain weight compared to now.”*

*“Argh! I thought it’s normal for a child to gain weight as he grows up, because it was not that bad. But I was frustrated that I had to always buy him clothes.”*

Dikeledi mentioned that Kamo eats at the mall after school and at weekends, but when he comes home he does not eat much. She tried to control his eating habits at the mall by talking to the people responsible for providing him with food there and requesting that they do not give him food anymore. However, this didn’t work as Kamo bought himself food when he got paid (for work he does at the mall: see section 6.4.2.6), and she gave in.

*“He doesn’t eat that much; even in the evenings he doesn’t eat much.”*

*“They also give him food. Where they sell chips they will give him, where they sell ice cream, those white people, give him ice cream.”*

*“I’ve gone to the shops and told them not to give him food, but he is not bothered by that, because he still uses the money they pay him.”*

*“Mhm, but he has stopped going there a lot.”*

Children with PWS may develop a wide range of food-related behaviours, such as running away from home to search for food in a wider area, which is potentially dangerous and difficult to manage (Honey, 2010; McCandless et al., 2011). Kamo may not be running away from home but he likes going to the mall. Going to the mall is not a problem; the problem is the possibility that he eats while he is there, and the primary caregiver cannot control his calorie intake.

*“I can’t really say it’s a problem. The problem is the obesity and the possibility that he eats when he is there.”*

According to Dikeledi, Kamo is aware that there are certain foods he is not supposed to eat, but he does not listen as he strongly believes that God gave him his weight.

Stubbornness, resistance to change, and a tendency to be very argumentative, are again reminiscent of the behaviour of a very young child, and can be linked to the person's emotional immaturity. These characteristics are observed in individuals with PWS (Ho & Dimitropoulos, 2010; McCandless et al., 2011). The primary caregiver has experienced these issues with Kamo especially when he is warned about weight gain.

*"Yes, he is, but he will tell you: 'It's my weight, leave me alone, stop telling me about my weight. God has given it to me; it's mine.'"*

Dikeledi's other complaint was that when she has left her juice in the refrigerator, Kamo drinks it. She currently does not lock up food because Kamo does not steal it. The manner in which she spoke about this came across as though it was not a persistent issue which was out of control. It was manageable.

*"No, I don't hide food."*

According to IPWSO (2013) control of food-related behaviours is complex, and centres on strategies to limit access to food such as consistent limit setting, locking cabinets and the refrigerator at home, limiting exposure to food, and avoidance of work environments with available food. Contrary to this, as stated by the primary caregiver, Kamo has unlimited food access, which is a serious concern given that he is gaining weight. However, the above-mentioned behaviour is reported to have improved since Kamo is no longer going to the mall. What is worrying, however, is that Kamo still has unlimited food access to food at home and presents with resistance and stubbornness. It may be difficult to control him in the future.

#### **6.4.1.11 Satiety**

Dikeledi implied that Kamo does not have satiety problems, because when he eats he gets full, and he is also satisfied with the small amount of food he gets; he even complains when he is given a bigger portion. The lack of satiety is also evident in that when Kamo has eaten at school and at the mall he gets full and then does not eat much or at all in the evening. This contradicts Honey's (2010) statement that the vast majority of individuals with PWS eat continuously and show no slowing down in their eating behaviour when faced with food.

*“He does not have satiety problems. He eats what you give him. He is satisfied with what you give him, and after eating he will put the dish away.”*

Being informed that he does not have satiety problems put me at ease because it means that even if Kamo has unlimited access to food, he has some control over his consumption.

#### **6.4.1.12 Weight control**

Dikeledi tries to control Kamo’s weight by giving him small meals. This is in line with suggestions made by IPWSO (2013) that it is imperative to begin good meal management and education at an early age in PWS individuals. This includes sticking to a strict schedule for meals and snacks, and also limiting portion sizes. On the other hand, Dikeledi does not seem to have total control over Kamo’s food intake while he is at school and at the mall.

*“A small portion, he knows. He will even complain when you give him a lot of food.”*

Dikeledi also gives Kamo healthy foods and believes that the growth hormone therapy is also helping Kamo with weight control.

*“This treatment helps, and I also try and not give him fatty foods and the other unhealthy foods.”*

Exercise is a very important factor in weight maintenance, and early establishment of a routine of regular daily physical activity, of at least 30 minutes, is strongly recommended (IPWSO, 2013). Kamo and Dikeledi jog every morning before he goes to school – but they took a break during the winter season because it was dark.

*“We run, but we stopped in winter, but now that it is summer we will start again.”*

Despite her efforts, Kamo’s weight was not controlled and this could be because he spends much time away from home and responds stubbornly when he is warned about possible weight gain. Whitman (1995) stated that behaviour management

must focus on preventing the opportunity for expression of such behaviours – rather than depending on the development of internal cognitive controls. The fact that Dikeledi is unable to control the environment, may prove to be a challenge in the future.

#### **6.4.1.13 Behaviour problems**

Dikeledi described Kamo as naughty, troublesome and “giving her a headache”, but she could not expand on these characteristics. She did not have tangible examples, which made me believe that the naughtiness she describes is generally observed and experienced in all children of Kamo’s age. What worried me was when Dikeledi reported that when she slaps or beats Kamo, in an attempt to discipline him, he slaps her back. She also complained that Kamo takes a long time when bathing, and that this causes conflict between them, and results in her beating him.

*“This child gives me a headache.”*

*“He is troublesome. I really don’t know what to say.”*

*“Kamo doesn’t listen, he doesn’t listen.”*

*“There is nothing that he does persistently that I would like him to stop doing.”*

*“[Pause] I really can’t pin-point; he is just naughty.”*

*“When you slap him, he slaps you back.”*

*“He wakes up at five o’clock to go to school, but at six o’clock you will find him still busy bathing.”*

Besides the above Dikeledi could not identify any other problem. She also accepted and acknowledged that Kamo does not have serious behavioural problems.

*“I really cannot remember anything else.”*

*“Kamo is really not that bad.”*

This suggested that Dikeledi is not experiencing most of the documented behavioural problems reported in individuals with Prader-Willi Syndrome and their caregivers.

Kamo seems to be presenting with milder behavioural problems. Based on the information gathered from the primary caregiver, Kamo seems to be presenting with stubbornness when it comes to warnings about possible weight gain. He also acts

aggressively when the primary caregiver beats him. These findings concur with IPWSO (2013) in that behavioural challenges range from very mild to very severe, and each individual can display differing behaviours.

#### **6.4.1.14    Independency**

Kamo seems to be quite independent. When he comes back from school he works at the mall, and with that income he buys himself cosmetics.

*“He used to bring money before. I think he no longer gets that much because he does not bring it anymore. When he has money, he buys himself perfumes and roll-ons.”*

#### **6.4.1.15    Adaptive functioning**

Kamo can bath himself and take care of his personal hygiene, but Dikeledi was concerned because he uses a lot of soap when bathing and takes a long time.

*“When he is bathing, he uses a lot of soap. He will apply so much soap on his towel – and I always wonder why.”*

#### **6.4.1.15    Social life**

Dikeledi mentioned that Kamo spends most of his time at the mall working as a patrol officer, and he is popular there with other workers and customers.

*“He works and they give him money. They know him well in the mall. Everyone knows him there – even people that shop there.”*

Kamo barely has time to play, but when he does so, he prefers to play with younger children and not his peers. This behaviour is discussed in the literature: because the social cognition of people with PWS may be impaired, most people with the syndrome have difficulties relating to their peer groups, and often prefer to be with older or younger groups (Whittington & Holland, 2010).

*“He does not have time for other children.”*  
*“He plays with the younger ones.”*

#### **6.4.1.16    Social support**

Dikeledi stated that nobody supports her, but she is used to it.

*“I don’t have time for that.”*

*“[Pause] what support? I am used to being on my own.”*

However, Kamo occasionally visits his aunt.

*“He occasionally visits my sister – but she stays far [away].”*

#### **6.4.1.17 Marital support**

Dikeledi’s husband was accepting of Kamo’s condition and supportive of her. Caldwell and Taylor (1988) indicated differing views on how the birth of the PWS child impacts on marriage. Some parents indicated it had no impact, while others mentioned that it strengthened their relationship as a result of mutual support – as was the case with this family.

#### **6.4.1.18 Government support**

Dikeledi receives an old-age grant and Kamo is a recipient of a disability grant.

#### **6.4.1.19 Future plans**

Dikeledi struggled to answer this question, and presented a pessimistic view. She could not imagine Kamo having a brighter future. She only worries about how she will manage to continue taking care of Kamo – as she is getting older and weaker.

*“Does he have a future? Ayi, Kamo! Ayi! No!”*

#### **6.4.1.20 Psychological support**

Dikeledi was worried about the fact that Kamo fights back and she needed assistance with that.

*“[Pause] It’s just this slapping!”*

#### **6.4.1.21 Prader-Willi Syndrome Association of South Africa (PWSSA)**

Dikeledi and Kamo are affiliated with the PWSSA. However, they do not benefit much from the organization. Dikeledi perceives the Annual General meeting as a

waste of time and money. This is because she attends, but leaves the meeting without having understood its contents or the documents as she cannot understand English and is illiterate. She therefore has to find someone to translate and to read it to her.

*“It’s been a while. As a matter of fact, I thought I wasn’t going to join them for the meeting this year, because while they [are] busy talking, I don’t hear a word of what they are saying.”*

*“I just go there to be an additional person, but it is not worth it.”*

This left me wondering whether the committee of the Association is aware of this sort of problem, and if they are trying to accommodate those who cannot understand English. Dikeledi believes they know of her need but are not doing anything to accommodate her.

*“They are aware, because Anna has been telling them, but they are not doing anything about it.”*

*“I don’t get information. I am always kept in the dark.”*

*“I normally ask Anna to tell me when they are done so that I can also stand up and go, but I don’t hear much; there are areas where I can understand a little, but mostly I don’t because I am not educated.”*

I found her narrative on this issue heart breaking, as she was part of the Association but her presence was not acknowledged. She spends money to catch several taxis to get to the meeting, and yet it does not benefit her in any way.

#### **6.4.2 Themes identified from the interview with Kamo**

The interview with Kamo was difficult to conduct, due to his speech difficulties. He struggled to express himself clearly, and did not elaborate on his answers. He gave one-word answers. The following themes were identified from our discussions:

##### **6.4.2.1 Diagnosis**

Kamo knew about his diagnosis and stated that his mother told him when he was still a child. As a result, he grew up with the knowledge.

*"It's an illness."*

*"I knew it when I was growing up."*

#### **6.4.2.2 Impact of PWS on his life**

Kamo mentioned that he is experiencing challenges with his weight, and that he wants to get rid of it. It was evident that the weight gain has impacted him a lot to the point that he suggested that the fat from his stomach be removed.

*"It's the weight."*

*"Cut the fat from my stomach."*

#### **6.4.2.3 Food-related problems**

Kamo admitted to eating a lot, and mentioned that he enjoys eating bread and fat cakes.

#### **6.4.2.4 Weight control**

Kamo is trying to lose weight by staying at home and not going to the mall anymore. He also used to exercise with his mother, but they have stopped, but he did not know the reason why they had stopped.

*"I stay home."*

*"My mother used to take me to the ground to run – but she stopped."*

#### **6.4.2.5 Social life**

Kamo reports spending most of his time working at the mall as a patrol officer. He mentioned that he has friends both at school and in his neighbourhood. They spend time talking and playing ball. He further stated that he enjoys having conversations with his friends.

#### **6.4.2.6 Employment**

Kamo works part-time at the mall as a patrol officer, and gets paid anything from R5 to R10 a session.

*"The manager of the mall ... gives me R10, sometimes R5, anything. I patrol everywhere."*



#### **6.4.2.7 Schooling**

Kamo mentioned that he enjoys going to school. Both the teachers and the other learners treat him well.

#### **6.4.2.8 Future plans**

Kamo reported that he would like to become a “traffic cop” when he grows up.

*“I want to be a traffic cop.”*

### **6.5 Interventions**

- ***Client-centred approach***

A client-centred approach was adopted with this family as stated earlier. According to Corey (2009) the client-centred approach of Carl Rogers emphasizes three core conditions: empathy, unconditional positive regard, and congruence. They are not formulated as skills to be acquired, but rather as personal attitudes or attributes experienced by the therapist, and are also communicated to the client in order for therapy to be successful. If therapists communicate these attitudes, the client will become less defensive and more open to themselves and their world, and they will behave in prosocial and constructive ways (Corey, 2009).

The key characteristic of empathy is understanding another person’s subjective reality, as they experience it at any given moment. This requires an orientation toward the client’s “frame of reference”. Accurate, empathic understanding implies that the therapist will sense the client’s feelings, as if they were their own, without becoming lost in those feelings (Corey, 2009). Unconditional, positive regard refers to the experiencing and offering of a consistently accepting, non-judgemental and valuing attitude toward a client. The caring is non-possessive and is not contaminated by evaluation or judgement of the client’s feelings, thoughts, and behaviour as good or bad. Congruence implies that the therapist is real, genuine, integrated, and authentic. The therapist openly expresses feelings, thoughts, reactions and attitudes that are present in the relationship with the client (Corey, 2009).

- ***Psycho-education***

According to Lukens and McFarlane (2004) psycho-education is one of the most effective evidence-based practices that has emerged in both clinical trials and community settings. It is a professionally delivered treatment modality that integrates and synergizes psychotherapeutic and educational interventions. Psycho-education reflects a more holistic and competent-based approach stressing health, collaboration, coping, and empowerment.

A psycho-education model therefore views the role of the psychological practitioner not in terms of abnormality, diagnosis, prescription, therapy and cure but rather in terms of goal-setting, skills teaching, satisfaction, or goal achievement (Authier, 1977).

Family psycho-education includes the teaching of coping strategies and problem-solving skills to families, friends, and/or caregivers to help them deal more effectively with the individual. The rationale for the psycho-education, is that it reduces distress, confusion, and anxiety within the family which may, in turn, help the individual to improve their lifestyle (Ahmed, 2004).

Because the primary caregiver is illiterate, does not understand English, and has forgotten most of the information she was given by the healthcare professionals during genetic counselling it was crucial to psycho-educate this family. First, the focus was on the importance of effective weight management, and the fact that if it is not managed effectively, it may predispose Kamo to numerous medical conditions, as discussed in the literature in Chapters one and two. During a follow-up intervention interview, the primary caregiver reported that Kamo is no longer going to the mall, and that his weight has since improved. However, she complained that occasionally Kamo gets stubborn and wants more food.

Secondly, the primary caregiver was psycho-educated on the cognitive functioning of an individual with PWS as described in Chapters one and two. The aim was to assist the primary caregiver to understand that beating Kamo does not serve its intended purpose, which is to teach Kamo right from wrong and to discipline him. Giving the primary caregiver information, assisted her to better understand Kamo's behaviour.

In the process, she was empowered as she got information in a language she understood, and for the first time she reported that she felt involved in her son's treatment plan. This further facilitated the discussion on different strategies of managing the undesired behaviour, which are described in the next section.

- ***Behaviour therapy***

According to Corey (2009) behaviour therapy practitioners focus on observable behaviour, current determinants of behaviour, learning experiences that promote behaviour change, tailoring treatment strategies, assessment, and evaluation. The aim of the session was to define the problem, identify what needed to be changed, assess the effectiveness or ineffectiveness of the current intervention, and devise intervention strategies. This is supported by Grant and Evans (1994) who advised that when facing a behavioural problem one of the first things is to identify what needs to be changed. They further emphasized that it is important for the behaviours to be clearly defined.

- ***Defining the problem***

Dikeledi struggled to define the problem but she described Kamo as naughty, troublesome, the fact that he does not listen, and gives her a headache.

- ***Identifying what needs to be changed***

Dikeledi requested that I assist her to stop Kamo from hitting her back. We had to identify incidents that possibly lead to her slapping him but she struggled to come up with practical examples. She could only recall that he drinks her juice from the fridge and that he takes a long time to bath.

- ***Assessing the effectiveness and ineffectiveness of the current intervention***

Beating Kamo was clearly ineffective.

- ***Anticipatory planning***

Good management means always anticipating tomorrow, next week, new situations, changed plans, and the like (Whitman, 1995). Based on the above the following were suggested:

- The behaviour needs to be managed by preventing it from happening, by hiding Dikeledi's juice, and putting it back in the fridge when Kamo has left for school or has gone to the mall. Since implementing this suggestion, they have not experienced any further problems.
- Dikeledi wakes Kamo up 30 minutes earlier, and every 10 minutes alerts him to the passing time. This strategy worked as Kamo would finish a few minutes before the transport arrived to take him to school.

Two weeks later, a follow-up session was done and the primary caregiver reported that she had stopped beating and slapping Kamo as a way of disciplining him. Two months later, the primary caregiver reported that since she had stopped beating Kamo, they had not experienced any problems.

Following these interventions, Dikeledi would occasionally contact me if she needed information on Kamo's behaviour, or if she required more information about PWS. This suggested that she needed this kind of assistance and support as she had nowhere else to go to source it.

Eleven months later, Dikeledi contacted me and an appointment was arranged for her to come see me at Dr. George Mukhari Academic Hospital. She came with Kamo, and reported that she was concerned about Kamo's behaviour as he was having occasional bouts of aggressive behaviour. During these episodes he broke a window and the gate and he chased a neighbour with a knife. This aggressive behaviour panicked me, and to manage these aggressive outbursts I referred Kamo to Paediatrics and Psychiatry. He was then prescribed Epilim, an anticonvulsant or mood stabiliser, in order to manage this behaviour. Dikeledi gave me feedback after two weeks, and Kamo had not had an additional aggressive episode, and he was also calmer.

Dikeledi also informed me that Kamo's appetite had increased – he was eating excessively and was stubborn when he was told to stop. When there is food left over

he will eat everything. This suggested to me that Kamo's satiety problems might only be developing now. Because the family does not have adequate space, I suggested that Dikeledi put a lock on the refrigerator, and buy a trunk where she can keep the dry ingredients stored and locked away. She further mentioned that Kamo defecates on himself and that she was embarrassed to tell me about this.

A month after initiating treatment Dikeledi reported that Kamo had not displayed any aggressive behaviour since starting the treatment. Kamo's weight had not improved; he was still obese. Dikeledi attributed the maintained weight to her inability to afford to buy the trunk and lock for the refrigerator, but recently she has been trying to manage Kamo's weight by cooking small meals and they consume it all to avoid having left-over food.

The socioeconomic circumstances of this family seem to contribute negatively in Dikeledi's attempts to try and manage Kamo's weight. The environmental modification required in managing food-related problems is not possible for this family because they do not have adequate space. Due to their family income they also buy food they can afford and not the food suggested by the dietician. This then demonstrates that the socioeconomic status has an effect on type of food-management approaches used.

## **6.6 Conclusion**

It is evident from the background information that Kamo presents with some of the features of PWS such as hypotonia, hypogonadism and cognitive impairment. There were no behavioural problems reported in early childhood, but in his early adolescence he occasionally responded stubbornly when warned about food and his weight. He initially did not display most of the behavioural problems mentioned in the literature, and this suggests that Kamo presents with milder behavioural problems. Kamo's presentation was initially not typical of the individual with PWS as presented in much of the literature. Instead, it seems to confirm the statement made by Ho and Dimitropoulos (2010) that PWS is a spectrum disorder, meaning that not all symptoms occur in everyone affected, and the symptoms may range from mild to severe. However, a few months later Kamo had bouts of aggression and had to be put on medication to control it.

It was initially intriguing that Kamo was reported to not have satiety problems. According to Goranson (2011) researchers strongly believe that the hypothalamus regulates appetite, metabolism, body temperature and mood in people with PWS. As discussed in the earlier case studies, it would be interesting to establish (possibly by using MRI imagery) if there are individual differences relating to satiety problems in the hypothalamus of individuals with PWS, or if it changes over time. However, eleven months later there was a significant increase in Kamo's appetite and he was eating excessively.

Although Kamo was on GHT, he was obese and had a short stature. Of great concern was the obesity and that the primary caregiver did not seem to have total control over his food intake when he is outside the home environment. The poor academic background of the primary caregiver seems to have contributed to how she has perceived her son's condition, and how she has been managing it.

The primary caregiver clearly lacks an adequate support system. Unfortunately, the PWSSA, which is meant to be part of Dikeledi's support system, has not met her needs. Being part of the research process was of significant benefit to the primary caregiver, as she could equip herself with information and manage Kamo better.

However, their socioeconomic status has a negative effect on type of food-management approaches used, which made it difficult for Dikeledi to control Kamo's weight. This considered, a longitudinal study may benefit this family in terms of implementing different interventions, evaluating their effectiveness over a longer period, and recording any changes in behaviour and development throughout their lives. This case also confirmed the effectiveness of a combination of psycho-education, behaviour management techniques, where anticipatory planning was implemented, as well as medication.

## **CHAPTER 7**

### **REBA'S STORY: "BUT NOW I AM LEFT ALONE"**

## **7.1 Introduction**

In this chapter I describe the lived experiences of Irene, the primary caregiver, and her son Reba, who has been diagnosed with PWS. First, I will highlight the devastating impact of the diagnosis on the parents. Secondly, I will demonstrate that caring for an individual with PWS is emotionally exhausting and time-consuming for the primary caregiver. Lastly, I will illustrate the difficulties experienced in managing behaviour problems, and how they also have a significant impact in the school environment. The findings are presented according to the topics discussed in the interviews and the themes which emerged. The statements are supported by italicised and indented quotes from the transcripts. The findings are integrated with the literature reviewed in chapters one and two.

Six sessions of approximately two hours each were conducted with this family. During the first session I interviewed Irene, and supported her with grief counselling because her husband, Victor, had died only a month previously. The second session was conducted with Reba in the presence of his mother, and further grief counselling was done. The third session focused on identifying the problems, assessing existing interventions, and devising additional interventions. The fourth session focused on getting feedback on the suggested interventions, and also grief counselling. The fifth session was used to gather collateral information from the deputy principal of the school Reba was attending. The last session was aimed at getting feedback on the interventions they were implementing, and there were further discussions with the deputy principal on interventions for behavioural problems. Telephonic contacts were conducted in between sessions, in order to get brief feedback. The first and second interviews were transcribed and translated, and process notes were written for the follow-up sessions using the voice recorder in order to preserve all the data. The interviews were conducted in Southern Sotho, Setswana and English.

## **7.2. Family background**

At the beginning of the data-collection process, this family was composed of Irene and Reba. Victor had passed away a month before I started collecting data. Irene was six months pregnant and she has since given birth to a baby boy. At present, the family comprises of the mother and her two children. They reside in a large,

beautiful home in a security complex in Centurion. They have a live-in nanny, Boitumelo. The family environment is conducive for living. Their family income is more than R20000 a month, and they own a car. They are Christians and attend the Centurion Christian Church every week, but took a break after Victor died. Their home languages are Setswana and English. Reba only speaks English. The reason for this is that he had speech difficulties, and it was recommended by the speech therapist that he should only learn one language. I therefore conducted my interviews with Reba in English only. The family is affiliated with the PWSSA.

### **7.3. The context**

Irene is a 34-year-old, widowed African female. She has an honours degree in accounting, is a chartered accountant, and is employed full-time as a lecturer. She was very welcoming and friendly. The fact that we were in the same age group helped me to develop a good rapport with her.

During the first session, Irene was very emotional as she narrated the circumstances around the husband's death and its impact on her life. A client-centred approach (this intervention is discussed in detail later) was adopted to allow her to express herself. During the interview, Irene also explored her experiences, in-depth, in terms of raising a child who had been diagnosed with PWS.

Reba is a 12-year-old, African male. He is currently a scholar in grade five. Irene transports him to and from school. He presented as friendly and warm when he fetched me from outside, but the interview with him was difficult until he spoke about his father. This process helped me to establish rapport with Reba. This was evident in that he requested a second session with me, as he felt I was the only person he could talk to about his father. In the follow-up session, Reba appeared to be more relaxed compared to the first time I saw him. He presented as friendly, spontaneous and cooperative. I reflected on the positive energy I observed in him.

Reba has observable dysmorphic features, observable gait difficulties, a short stature, small feet, his nails were bitten, and he had speech difficulties. His speech was unclear but audible in the beginning but during the course of the interview he spoke in a soft, inaudible tone. He struggled to pronounce some words. According to



Lewis (2006) speech difficulties include poor speech-sound development, which includes errors due to poor motor abilities associated with the production of speech-sounds and also errors in applying linguistic rules to combine sounds to form words.

Reba had a fairer skin than his mother. This concurs with the observation of Butler et al. (2006) who stated that PWS individuals may present with hypo-pigmentation. Reba is not a recipient of a disability grant, because his mother was not aware that he qualified for one. Information was given to them on different types of disability grants and also the rationale for a disability grant. Irene will independently start the application process.

## **7.4 Lived experiences**

The lived experiences of Irene and Reba were explored by conducting semi-structured interviews with them. From the transcribed interviews, certain themes were identified, which are discussed in the following sections.

### **7.4.1 Themes identified from interviews with Irene**

In the interview with Irene, the following themes became evident:

#### **7.4.1.1 Pre- and post-delivery**

Irene's pregnancy was full term. During her pregnancy she had decreased foetal movements, but she didn't know the cause of this. This concurs with the views of Cassidy and Driscoll (2009), Cassidy et al. (2012), Ho and Dimitropoulos (2010) and Hurren and Flack (2016) who stated that in individuals with PWS hypotonia is prenatal in onset, and usually manifests as decreased foetal movement. Irene mentioned that she gave birth in a private hospital.

*“Even while you are pregnant, they are not active and they can stay in one position forever. Even for more than an hour – so now I know because I have reference. At the time I didn't know.”*

She gave birth through caesarean section because the doctor was worried about the position of the umbilical cord which would entangle the baby. They first induced labour but this was unsuccessful and then a caesarean section was done. Following

the delivery, Reba had breathing problems and had to be admitted to the intensive care unit (ICU).

*“He didn’t breathe on his own, they had to take him to ICU.”*

Reba was floppy, he was also fed through a feeding tube, and his genitals were under-developed which is what caused the paediatrician to investigate. Under-developed sex organs such as undescended testes, a small phallus or small clitoris (hypogonadism) are another indicator of PWS (McCandless et al., 2011).

Reba and Irene were admitted to hospital for two weeks and her husband came to visit them regularly. By the time they were discharged, Reba could breathe on his own and feed with a bottle. Irene and Victor sensed that there was something wrong with the baby, because, in addition to breathing problems and floppiness, Reba was not crying. He was sleeping excessively, and they had to wake him up for a feed. These issues have also been identified as other characteristics of hypotonia. Hurren and Flack (2016) and McCandless et al. (2011) have stated that PWS should be considered in any infant with significant poor muscle tone (hypotonia) particularly in settings of poor feeding and reduced spontaneous arousal for feeding.

*“Because he was floppy, his genitals had not developed. Reba didn’t cry at all and he slept throughout.”*

*“He was fed through the tube.”*

#### **7.4.1.2 Diagnosis**

Irene and Victor got the diagnosis a week after they were discharged from the hospital. They were devastated by this, because the doctor informed them that Reba would be retarded. They had hopes and dreams for Reba’s future and it was very disappointing that he would not live up to those dreams. According to Caldwell and Taylor (1988) PWS children had a poor prognosis with a high probability of having significant cognitive limitations.

*“But you have dreams – so now you have to terminate those dreams, immediately, and think something completely different that your mind can’t even design a plan, so you work from having some plan to now having to go on day-by-day.”*

*“When they tell you, the confirmation is devastating and you have so many questions.”*

They found it very difficult to cope with the diagnosis. Irene’s studies were also interrupted as she had to take time off after Reba’s birth. They were not prepared for the diagnosis and even wished that he could die.

*“Eyi! Difficult yho! I think we had so many ... well both of us, we were the same at some point. We thought it would be better if he dies.”*

Irene reported that when Reba was initially diagnosed, the fluorescence *in situ* hybridization (FISH) test was used to confirm the diagnosis after birth. McCandless et al. (2011) stated that testing for PWS should begin with DNA methylation analysis, and if the methylation analysis is consistent with PWS the FISH is used to detect a deletion on chromosome 15.

The family recently had a chromosome test done, which the husband insisted on having, and the results came back as undetectable because the deletion is milder. Irene believes that Reba’s features are not prominent compared to those of other individuals with PWS except for his height. Miller et al. (2010) and Driscoll, Miller, Schwartz and Cassidy (2016) state that chromosome microarray (CMA) testing is a relatively new test, but is becoming the test of choice for detecting most chromosome deletions even many small “atypical” sizes and it will also detect many cases of uniparental disomy 15 (UPD), which is another genetic subtype of PWS.

*“He told the doctor, ‘I want to do a chromosome test to check.’”*  
*“You can tell, because he is quite short.”*

The diagnosis had a significant impact on Irene and Victor, to the point where Victor researched family members to try and understand the genetic origins of the syndrome. They did a family tree to check who in the family had had the syndrome, and they even had siblings tested. They needed answers as to why their child had PWS.

*“We did a lot of research checking family members.”*

#### **7.4.1.3 Feeding problems**

Assisted feeding through special feeding techniques including a feeding tube, special nipples and/or gavage feeding with increased feeding times are necessary usually during the first weeks to months of life, in order to ensure adequate nutrition and avoiding failure to thrive (Cassidy & Driscoll, 2009; Cassidy et.al., 2012; Driscoll et al., 2016; Ho & Dimitropoulos, 2010). Reba had a feeding tube for two weeks, and after two weeks he was bottle fed. Feeding him was a challenge as he slept during feeds. Victor came up with his own technique of how to keep Reba awake during feeds – by tapping the bottom of the bottle. This helped Reba to remain alert until he finished feeding.

*“He used to tap the bottle at the bottom the whole time.”*

#### **7.4.1.4 Milestone developments**

As Reba’s speech was delayed, he had to see a speech therapist, who suggested they only speak English to him – to assist with speech development. As a result, he cannot speak his mother tongue.

*“He spoke [for the first time] at two years six months to three years.”*

Cassidy and Driscoll (2009), Driscoll et al. (2016) and Hurren and Flack (2016) all stated that children with PWS experience delayed milestones including gross motor and language delays. Early milestones are reached, on average, at double the normal age – for example, sitting at 12 months, walking at 24 months, and speaking words at 2 years, as with Reba.

#### **7.4.1.5 Medical problems**

According to Verrotti et. al (2014) the frequency of epilepsy in PWS patients is higher than in the general population, and ranges from 4% to 26%. Epilepsy in PWS is usually responsive to anti-epileptic monotherapy, with rapid seizure control and a good outcome. Reba had febrile seizures commencing at eight months and he was prescribed Epilim until the age of two. Epilim is used to treat absence or petit mal seizures (the epileptic episode may go unrecognized, because the characteristic motor or sensory manifestations of epilepsy may be absent or so light that they do not arouse suspicion), myclonic generalized tonic-clonic akinetic symptoms (exhibit

classic symptoms of loss of consciousness, generalized tonic-clonic movements of limbs, tongue biting, and incontinence), and partial seizures (simple without alteration in consciousness or complex with an alteration in consciousness) (Sadock & Sadock, 2003).

Irene mentioned that from the paternal side of the family, most family members were epileptic, including Reba's father, but it was often observed after a specific incident which usually had an onset in adulthood. Therefore, it was associated with a traumatic incident or a stressful life event. Reba's seizures were associated with fever. Irene also reported that Reba's milk teeth did not fall out like in his peers. He had to be taken to theatre to have them extracted.

#### **7.4.1.6 Satiety and food-related problems**

The difficulties with feeding suddenly changed to excessive eating. King (2008) reported that two separate and distinct eating disorders are noted: initial feeding difficulties and failure to thrive, and then, later, overeating as in this case.

When they started Reba on solid foods at seven months, Irene realized that Reba was able to eat more than other infants. According to Butler et al. (2006), Honey (2010) and King (2008) in the second phase the disorder continues with hyperphagia (excessive appetite for food), which seemingly begins by eighteen months to two years of age. However, Irene disagrees, and believes that by the age of two it is more observable, because the children start seeking food themselves. However, she strongly believes that individuals with PWS are born with satiety problems.

Reba has an excessive appetite which he cannot control. As a result, once he starts eating he cannot stop. This supports Honey's (2010) statement that the vast majority of individuals with PWS eat continuously and show no slowing down in their eating behaviour when faced with food.

*"At seven months he had started, he could never stop."*

*“No, it’s wrong because they say two, it’s not that. The problem is that by that time they are the ones who seek food. They are born with it, someone would stop at some point – but he will eat the whole day.”*

Irene described Reba as manipulative when he wants food. He will not ask for food directly, but comes up with ways to convince you to give him food.

*“He can come up with ways. For instance, if he doesn’t know you and he finds you here and you probably have a packet of chips, he will talk to you and ask you: ‘do you like chips?’, and maybe he will say ‘I like them and stuff’, and will ask you: ‘how many do you buy?’ That’s how manipulative he is – even with neighbours, I was surprised. My neighbour tells me that he often visits and she gives him sweets. When I ask if he asks for the sweets, she said: ‘no he is such a nice boy, I end up giving him.’”*

Irene finds it difficult to educate her extended family, friends, and neighbours about Reba’s food-related problems, because she believes people often reward children with food and do not comprehend why Reba cannot have food outside his diet schedule. She believes that the only people who seem to understand are the parents and professionals.

*“No, they wouldn’t understand, so they have to give him whatever, so Reba would look sad and they feel sorry for him [laughter]. They will say: ‘he can have an apple’, even when you explain to them, that an apple for him is the same as having a full meal, when he has that apple – where is he going to work it off?”*

The literature recommends that relatives and social contacts must be educated that “sneaking” food to the child with PWS is not an appropriate method of demonstrating affection, and undermines the child’s nutritional regimen and sense of wellbeing (IPWSO, 2013). However, this has proven to be a struggle for this family, as stated above, and the major challenge for Irene is to convince Boitumelo to refrain from offering Reba food, snacks or treats outside his diet. Unfortunately, Boitumelo also forms a coalition with him, as she gives him food in her room despite Irene’s strict instructions not to do so.

*“Here I am planning meals for him but I don’t know how many calories they exceed and in a week – we have always battled with this.”*

Irene suggested that I talk to the nanny as it might make a difference if she speaks to a professional. She was even considering getting Reba an *au pair*, as she is worried about the nanny not adhering to the diet plan. I was really concerned about the fact that the nanny demonstrated a lack of consistency and could not adhere to the schedules as she was supposed to be assisting Irene. Another concern for me was that Reba could develop food-related complications like obesity or diabetes if the nanny continued with non-compliance. Cassidy and Discroll (2009) found that if intake is not controlled externally, obesity results from these behaviours and combines with a low metabolic rate and decreased activity level. Complications of obesity are the major causes of morbidity and mortality: cardio-respiratory insufficiency, obstructive sleep apnea, thrombophlebitis (vein inflammation), and chronic leg oedema (swelling).

Irene reported that they had introduced a culture of eating in front of Reba and eating differently from him because she believes that it is something he has to deal with when he is at school, at a mall, and also other places where people eat together. She believes that Reba needs to be able to control himself at home if he is to cope with what happens at school. What this family has chosen to do has been discouraged in PWSUK (n.d.) which suggests that others should not eat in front of the person with PWS.

*“No, aha! We’ve always said as well that he has to get used to it.”*

Reba finds ways to get his hands-on money in order to buy food or snacks at the school’s tuck shop. For example, he sold the puzzles and sweets he got from his grandfather to people who were visiting the house, and made R40 which he used to buy snacks at school. Irene could not do anything about it.

*“Mhm, mhm, how do you completely control that part with complete strangers – because I can’t explain to people.”*

Whitman (1995), McCandless et al. (2011) and IPSWO (2013) state that the physical living environment must be designed so that food delivery, food access, food preparation and food disposal, are all managed in a fashion that is neither visible nor

accessible to the person with PWS. This usually implies locked cabinets, pantries, refrigerators, and, ideally, totally locked kitchens and there should be close supervision at all ages. One should also limit exposure that makes the child think about food.

The family is struggling to ensure that Reba has restricted access to food. In the past they tried locking the refrigerator but it did not work for them because he would sneak into their bedroom and steal the key while they were sleeping, and sometimes they would misplace the key and it then became an inconvenience for them. These issues echo Gourash et al.'s (2006) findings that individuals may pick and break locks or steal keys in order to enter a locked kitchen. They may also have violent outbursts or aggression related to food acquisition or to attempts to set appropriate limits. Families often find themselves in a situation which is spiralling out of control.

They even came up with a strategy to set the alarm differently to prevent him going into the kitchen at night to steal food but he used to move so quickly that they would find him in his bed pretending to be asleep. As a result, they were continuously monitoring him, locking the refrigerator, and hiding the key which was exhausting for them. They had to choose between monitoring the key or monitoring the person, and they decided on monitoring the person. Currently, they do not lock, but rather monitor Reba by ensuring that he is never left alone. The only way he has access to food is if he steals the food on his own.

*"No, we don't lock – we tried it but it didn't work."*

*"If he wants to steal, yes, but that's the only way he will go and get food."*

*"We would rather micro-manage him."*

#### **7.4.1.7 Weight control**

Irene adheres to a strict diet and dietary plan for the day, and she prepares Reba for what he will have, and tells him in advance if they are going to make changes to his diet. She gives him a lunch box with calculated calories when he goes to school. She does not buy sweets and chips, and, as a result, he does not have a supply at home.



According to Irene, Reba's weight is controlled compared to other children with PWS, and she believes he is doing exceptionally well given that he is currently not taking growth hormone therapy (GHT). But since the father's passing Reba has gained a lot of weight because he has had easy access to food, as no one was monitoring his intake as the family was focusing on funeral arrangements. Irene mentioned that because of his low muscle tone and slow metabolic rate, it is easier for Reba to gain weight. It is worrying that Reba is gaining weight and there is no adequate control around his food access.

*"Because of the funeral, he has gained a lot of weight but without growth hormone and everything else we've managed to really control his weight, so he is supposed to be bigger and obese – like extremely obese."*

The parents used to encourage Reba to exercise on a treadmill, but he refused to do so. Then they bought him a bicycle so that he could use it to exercise but he does not ride it. This concurs with Butler et al. (2006) who established that PWS children are less physically active than their peers. IPWSO (2013) emphasized that exercise is a very important factor in weight maintenance, and early establishment of a routine of regular daily physical activity, of at least 30 minutes, is strongly recommended. However, this is not happening with Reba, which may contribute to the difficulties with his weight control.

#### **7.4.1.8 Growth hormone therapy (GHT)**

Irene wished that she could have given birth in a government hospital in terms of receiving GHT. They used to fund the treatment themselves because the medical aid does not cover it but it was too costly at R3300 a month. Subsequently they had to stop it, and this compromised his growth.

*"I can't really afford it, it's too much."*

*"So, if Reba was normal height, at least [his] body mass index would be ... yah because if you are tall, you don't look too fat."*

Goranson (2011) reported that only after 1990 did scientific studies indicate that the use of human growth hormones was beneficial for children born with PWS. While some children with PWS got treatment, others were denied treatment because it could not be proven that they had a growth hormone deficiency. Even if the doctor

prescribed it, the family health insurance plan typically refused to cover the cost, because it was considered an experimental treatment for children with PWS. For these families, because of the cost of medication the promise of a better life for their children was out of reach. It is shocking to find out from this family that a decade later, their medical aid still does not recognize GHT as a treatment for PWS. Subsequently, Reba's growth was further compromised.

#### **7.4.1.9 Impact of PWS on Irene**

Whittington and Holland (2010) stated that 70% of mothers of children with PWS experience high levels of stress, and need psychological counselling. This was the case with Irene who believes that PWS is one of the most difficult conditions to deal with – because she constantly has to manage her son. She is overwhelmed by this experience and needs help.

*“We have to constantly manage, [and] when I say constantly I mean every day, every minute, yes you can imagine, you don't rest. It's stressful because its constant management – it is not like you can afford not to have a nanny.”*

Irene was overwhelmed about the thought of giving birth to her second born as she was concerned about whether she would cope. She spoke in a “stuck” and despairing manner as she saw no way out for her.

*“And I have another baby coming, sometimes after death I was like ‘why didn't he go with Reba?’ I know it's a bad thing to say, but ... yah, at least if he had gone with Reba, then I wouldn't have so much.”*  
*“Yes, it feels like a burden, yes, because I have him for the rest of my life.”*

#### **7.4.1.10 Schooling**

Reba was first enrolled in a Montessori school and then moved to Raslouw Academy. The different teaching methods of these schools meant that Reba had to repeat grade one. Irene was not sure about the current school's speciality. She described it as a special school which offers matric, that caters for children with attention deficit hyperactivity disorder, dyslexia, and pupils who are of average intelligence. Reba is currently doing grade five. Regardless of the IQ score, however,

most people with PWS have multiple, severe, learning disabilities, and poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010). This appears to be the case with Reba, as he attends Raslouw Academy, which is actually a remedial school

*“When we moved him from Montessori to this school they said, no, they can’t take him directly from Montessori because it is different.”*

When Reba was admitted to the school, the parents informed the teachers about PWS, food-related problems and behavioural problems. However, they found it difficult to explain what the condition entailed to the teachers and it was also difficult for them to understand. They then thought of getting a professional to explain things to the teachers, and then arranged with Dr. Duncan who is a paediatrician and a specialist in genetics, to do a three-hour workshop at the school.

*“We had to organise because it was just difficult, because we tried to explain, looks like they were not getting it, so we thought if we get someone, a professional, to come and do the workshop ... Dr. Duncan went there, to do the workshop.”*

Reba is struggling to cope at school because he presents with behaviour problems. He is stubborn, steals, and tends to have uncontrollable temper tantrums. The parents are often called to school to fetch him. When his outbursts are frequent Irene often takes Reba to the doctor, who gives him time off from school or persuades the principal to give Reba a few days off.

Due to behavioural problems, Reba had to be home schooled at the beginning of 2016, until his father’s death later that year. The home schooling provided them with a break from problems at school, and put a stop to the food problems. Reba liked the one-on-one attention. Irene would have continued with home schooling if her husband had not died. Now she is considering enrolling Reba back at the Montessori school as they do not have a tuck shop.

#### **7.4.1.11 Decision to have another baby**

Irene emphasized that it was a well-considered decision to have another baby. After a long time of dealing with the fear of having another child, they finally found the courage to try again. It was difficult for people around them to understand why they

were not having another child because they never told them about Reba's syndrome. It is telling that they kept Reba's condition a secret because their friends would then not understand the anxiety of possibly having a second child with PWS.

*"It took a lot of faith to even get to a point where you say – 'let me, let's try, maybe this time around we could be lucky.'"*

They had to go for genetic counselling and had an amniocentesis to make sure that the baby did not have any genetic disorders. The amniocentesis results were negative. Being pregnant was very difficult for Irene without her husband's support. She feels she might not be able to cope with both Reba and the baby, and was actually thinking of asking her mother to help raise the baby. She is worried about the impact this will have on her mental state, and she is worried about post-partum depression.

*"I don't know if I will be in a state to raise the baby ... I don't know, I am worried I would have post-whatever."*

#### **7.4.1.12 Siblings**

Reba is aware that Irene is having a baby and has always wanted a sibling. He is even waiting impatiently for her belly to grow larger. Irene has involved Reba in the process by taking him to her ultrasound scan (sonar) appointments.

*"So, he is excited – he has always wanted a brother or sister."*

#### **7.4.1.13 Social life**

Reba struggles to socialize and Irene attributes this to the fact that Reba is stubborn and controlling. He does not play with his peers, and prefers to play with much younger children or with older people. She perceives this choice to be manipulative in that he can control the younger ones and the older ones will allow him to have his way. This relates to Whittington and Holland's (2010) findings that because social cognition may also be impaired, most PWS people have difficulties relating to their peer groups and often prefer to be with older or younger groups:

*"He struggles with socializing, because they are so controlling and so stubborn."*

*“He will prefer to play with extremely young or, yah, or extremely old.”*

#### **7.4.1.14 Social support**

Irene finds it very difficult to tell people about Reba’s condition. As a result, only a few people knew about it until recently. The reason for this is that she does not want people to treat Reba differently or to offer her sympathy. This further deprives her of social support. She believes that people will never understand because they do not have a lived experience, and it becomes difficult to explain.

*“I don’t think you ever do, like you’re not living with this thing, you don’t ... People don’t comprehend, I don’t know how to explain it.”*

Irene currently receives support from her friends and family members but they do not stay close by, and she does get no support from the husband’s family as their relationship changed after her husband’s death.

*“I have to manage all by myself.”*

#### **7.4.1.15 Marital support**

Irene was married to Victor for 13 years; they got married when she was 20 years old. She found her identity in her marriage, grew in her marriage, and they did everything together. Therefore, losing her husband had an enormous impact on her, her life, and she is really struggling to cope.

*“I don’t know, I don’t have an identity.”*

She mentioned that they never had a difficult marriage. Even Reba’s diagnosis and living with Reba did not impact negatively on their marriage. Instead, it assisted them to become closer, as they relied on each other and were each other’s support system. This concurs with Caldwell and Taylor (1988) who found that some parents indicated the PWS child had no impact, while others mentioned that it strengthened their relationship as a result of mutual support.

*“We trusted each other about Reba’s condition and stuff. You knew this is the only person you could depend on. I think that’s also something that brought us close, because we have to rely on each other.”*

For the first time following her husband's death, Irene had to disclose Reba's condition to many people. Before this, she did not think it necessary, because they were there for each other, and when they experienced problems related to Reba's condition together they found a way to resolve it. They even took a decision not to join the PWSSA, because they wanted to have their own subjective experience that was not contaminated by others' opinions. They only joined the PWSSA in 2015.

Irene feels desolate without her late husband's support, because he was always there to give her advice.

*"It will be very difficult – at least I had someone I could talk to."*

#### **7.4.1.16 Behaviour problems**

Butler et al. (2006) and Cassidy and Driscoll (2009) established that lying, stealing, temper tantrums, stubbornness, controlling and manipulative behaviour, compulsive-like behaviours and aggressive behaviour, are all common during the childhood years in PWS and continue into adolescence and adulthood. Children with PWS do not easily agree to anything, are resistant to new ideas and experiences, and are more dependent than typically developing children. Another identified aspect is lack of emotional control in children and adults with PWS. This could be attributed to the dysfunctional hypothalamus, which controls hormonal functions and emotional stability.

Cassidy and Driscoll (2009) and Whittington and Holland (2010) found that parents are divided on which of the above stated phenotypic characteristic is most stressful for them but most cite the eating behaviour or the obsessive-compulsive behaviours, depending on which are predominant in their child's behaviour. Closely related to these two characteristics is the problem of temper outbursts.

For this particular family, both kinds of behaviours have been problematic. Irene mentioned that Reba is stubborn, becomes irritable easily, is resistant to change, does not want to be told what to do, steals, is manipulative, has anger outbursts, and

has obsessive-compulsive behaviours. His obsessions last for a long period. His first obsession was about cars, and recently his obsession has been around Irene's medication and the funeral.

*"A lot of things like stubbornness, being easily irritable, having obsessive-compulsive disorder."*

*"So, anything relating to cars, he would obsess about everything, even conversations would be about cars, the whole conversation for almost two hours, it could be about that."*

*"I am now taking medication for pregnancy, those pregnancy vitamins, so it has to be taken at a certain time, and when he decides this is when it will be taken, yho! he gets obsessive about it!"*

The stubbornness and anger outbursts they experience with Reba were noticeable when he started crèche around the age of four or five. These characteristics also impacted on his progress at school as he would refuse to take instructions from the teacher. According to Cassidy and Driscoll (2009) it has been observed that characteristic behavioural patterns begin in early childhood, in 70-90% of affected individuals.

*"You can never tell him anything, he knows everything. There is nothing on this earth that he doesn't know. It doesn't matter what it is, for example at school when you say 'a cat is not spelt with a K it's spelt with a C', he will say 'no' and that time he will spell it the way he wants. When he finally decides to change it, he will. It's one of the things that his teachers are struggling with."*

Reba also does not comply with any instruction, defies authority, and will do everything in his power to ensure that whatever punishment you have for him you will not get to implement.

*"In everything he will make sure he wins and it will leave you feeling powerless."*

Reba tends to have outbursts that would last approximately 20 minutes. He says negative things when he is angry, will run, hit something, and sometimes he hurts himself. It appears as though he has no control, and the anger takes over. Together with her husband they were, and Irene still is battling with managing the anger

outbursts and stubbornness. She observed that what triggers the outbursts is mostly being told what to do. He also gets easily irritable when he is not in control because he likes to have control.

*“Because anger is one of the biggest ones, he gets obsessively angry, like uncontrollably angry ... ‘I don’t like you, I hate you’, all those things, ‘you don’t talk to me’. If it’s the aunt, ‘you are no longer my aunt.’”*

Irene also experiences stealing problems with Reba which are often related to food. She experiences the problem mostly when Reba is at school because they have a tuck shop there and that causes him to steal. Reba steals from her, the nanny, her siblings, the school teachers, and anywhere else where he can get money.

*“Because at school they have a tuck shop, so now he steals money and buys at school – which is one of the problems we have.”*

Irene described Reba’s behaviour as erratic. She feels over the years that his behaviour differed from that expected of Prader-Willi Syndrome except for stealing, where he steals money to buy food.

*“It’s like a different species of behaviour.”*

Irene also informed me that when Reba is upset, he defecates on himself and will urinate around the school naked while other school children laugh at him. He defecates on himself about twice a week.

One can therefore not under-estimate the significant impact this behaviour may have on the family. This was evident in how Irene spoke about this topic in an overwhelmed manner. It sounded like she and her husband had struggled with a lot of behavioural problems and needed answers or solutions. The parent’s concern was that behavioural problems were not given attention. When they attended their first PWSSA AGM, they were disappointed by the fact that there was only a dietician addressing people, but that behavioural problems were not given any attention. They felt there was adequate research and literature on diet, but only limited information on behaviour management.



*“His problem was not more on eating; his problem was more behaviour. They will have a dietician, but they will never have someone who talks about behaviour and addresses behaviour.”*

#### **7.4.1.17 Treatment received**

The family are currently consulting with a neurologist who is prescribing Concerta (methylphenidate) for Reba to help manage his behavioural problems. According to Sadock and Sadock (2003) Concerta is a central nervous system stimulant. It is used to treat attention deficit hyperactivity disorder and has been shown to have significant efficacy, and is safe. Boyle's (1997) findings suggest that this is the final class of drugs used for PW patients, and the medications may well be helpful for PW patients who have attention difficulties. It is interesting to learn from this family that a medication meant to treat attention deficit hyperactivity disorder, can help manage the range of behavioural problems Reba was presenting with.

During the first two to three months of Concerta treatment, they observed a significant improvement in Reba's behaviour. The medication also decreased his appetite, but after that his behaviour deteriorated, but the doctor assured them that the medication was working although it wouldn't completely eliminate the unacceptable behaviour. His Concerta dosage has since been increased from 18 mg to 27 mg. Unfortunately, it seems as if the effect of the medication has diminished with time.

*“It is not possible, it's not possible, it will not eliminate behavioural problems. It just helps you manage them – but it won't eliminate them.”*

The previous school principal who was a psychologist suggested in 2014 that Reba may be suffering from epilepsy, following an incident when she observed Reba appear to have a blank stare and to be unconscious for a few seconds while they were travelling in a car together. Irene and Victor then took Reba to the neurologist for an electroencephalogram scan (EEG) and a magnetic resonance imaging scan (MRI) but the results were normal.

#### **7.4.1.18 Routine and structure**

Irene reported that they have a routine and structure in place, but Reba never follows through with it. He does things the way he wants to, and when he wants to. If you remind him he will refuse to do what he is meant to do. He likes being in control and if you give him instructions, he gets more irritable and stubborn.

*“We try to have a routine, but the thing is when it comes to routine, he does it the way he wants to do it.”*

*“He likes to control, he even wants to control me.”*

#### **7.4.1.19 The nanny**

Irene reported that they have a live-in nanny, Boitumelo, who takes care of Reba during the day. Boitumelo struggles to get Reba to adhere to routine, because Reba disrespects her. She sometimes gives him food outside his routine because she feels sorry for him. She also tends to be inconsistent in her discipline.

*“For instance, if she is eating chips, she would give him [some].”*

He has a daily planned meal with calculated calories. Irene had to explain to Boitumelo the consequences of him over-eating, but in the beginning, she struggled to understand, so she gave her examples of extreme consequences in order for her to realize the seriousness of this condition.

*“My nanny in the first year struggled to understand. I had to explain, do shock therapy and show her, scare her that he is going to die ... they could die, there’s some who’ve had stomach, yah ruptures.”*

Irene further mentioned that Reba does not treat Boitumelo with the same amount of respect as he treats her. When Boitumelo gives Reba instructions, he will go and sit outside and starts shouting at the neighbours or say:

*“No! You not my mother, you get paid to do this job and they can fire you and get a new person.”*

#### **7.4.1.20 Government support**

Reba is currently not receiving a disability grant, because Irene was not aware that she could apply for one.

*“I didn’t know, there is a lot of things I don’t know ... I thought you only get old age grants.”*

#### **7.4.1.21 Future plans**

Irene and Victor prepared themselves for a possibility that they may have to live with Reba for the rest of their lives. Besides that, Irene does not have any future plans for Reba because she does not want to be disappointed. She only focuses on what he has to achieve in the given year; she only has short-term goals for him.

*“I don’t know how things will turn out, we had always planned for a worst-case scenario we might have to live with him, uh, until we die, because we don’t know if he will be completely independent. Even if he finishes matric there is no way that he will be independent around food ... I don’t know where he will end up ... I’d rather be happily surprised than think he will do this, so we work on what he achieves for that year – so for this year, it is to pass grade 5.”*

#### **7.4.1.22 Knowledge about other PWSSA members**

It seems that Irene communicates with other members of the PWSSA, as she knew that Makhotso’s daughter Tshepiso (discussed in Chapter 5), was receiving GHT from a government hospital – which is why she mentioned that she should have also given birth in a government hospital. She also mentioned that Makhotso is not experiencing serious behavioural problems with her child, only problems related to food which, according to her, is normal expected PWS behaviour. Furthermore, when she talks to Makhotso, she feels that she cannot relate to what she (Irene) is going through.

*“She says they get it there because that is where she gave birth, so we were trying last year to get there but it didn’t work because they can’t service you if you are not in that, what do you call it? ... in that region.”*

*“It does not help talking to Makhotso – she cannot relate to what I am going through.”*

I found it interesting that these two individuals both had children diagnosed with PWS but were having such different experiences. She also mentioned that she and Makhotso had one thing in common, which was the difficulty in deciding whether to have another child.

*“But, it’s not only me, I mean Makhotso, they also have one child. They were saying the same thing, that it is difficult.”*

#### **7.4.2 Themes identified from interviews with Reba**

The first interview with Reba was difficult to conduct as his speech was unclear: he spoke in a mumbled, low-tone voice. At times he refused to answer certain questions posed to him or he gave one-word answers and responded with non-verbal cues. He was either playing with his phone or skin picking when the questions were directed at his mother.

Grief counselling was initiated when Reba spoke about his father. For the first time in the interview he was able to express his feelings and became very emotional. I remained client-centred at this stage, and allowed Reba the space to express himself, and also mobilized him by providing him with empathy. His feelings were normalized and I prepared him for likely triggers. The caregiver also became emotional, and I used the space to facilitate a sense of cohesion.

Most information was gathered in the fourth session, when I was meeting Reba for the second time. He was at this time cooperative, friendly, and answered and expanded on questions posed to him. He even gave some information voluntarily. This could be attributed to the rapport established with him during the grief-counselling process. The following themes were identified from our discussions:

##### **7.4.2.1 Diagnosis**

Reba did not know about his diagnosis, as his parents have never told him. I assumed he knew and without knowing, I practically told him and that left me feeling uncomfortable. Irene indicated that Reba only knew that he had certain symptoms. Although it was clear that Irene and Victor preferred to be discreet about Reba’s diagnosis, I did not realize that it included Reba himself.

When I asked Reba about his symptoms, he said he did not know, until the primary caregiver intervened and then he finally agreed that he had satiety problems. The following extract from the transcript illustrates the stubbornness, perseveration and

argumentativeness Reba was presenting with throughout the first interview. It was beneficial that the primary caregiver was there otherwise I would not have sourced information from Reba:

**Molelekeng:** *Ok, do you know what your symptoms are?*

**Reba:** *Uh?*

**Molelekeng:** *Do you know what your symptoms are?*

**Reba:** *I don't know.*

**Molelekeng:** *What are you struggling with?*

**Reba:** *I don't know.*

**Molelekeng:** *On a day-to-day basis.*

**Reba:** *I don't know.*

**Irene:** *Tell her – you know.*

**Reba:** *I don't know.*

**Irene:** *You know them.*

**Reba:** *I don't.*

**Molelekeng:** *Maybe mom, you can tell me some of the symptoms and he will remember.*

**Irene:** *What about eating?*

**Reba:** *Mmmmmm.*

**Molelekeng:** *Is there a problem with eating?*

**Reba:** *Mhm?*

**Molelekeng:** *Do you have a problem with eating?*

**Reba:** *No.*

**Irene:** *No, tell the truth ... I mean.*

**Reba:** *Mhm! [Shrugging his shoulders]*

**Molelekeng:** *You don't know?*

**Irene:** *So, if I get stories from school it's not a problem?*

**Reba:** *[Shakes his head]*

**Irene:** *For you, It's not a problem,*

**Reba:** *[Shakes his head]*

**Irene:** *If you've stolen something from school, it's not a problem?*

**Reba:** *Mmmh.*

**Irene:** *How is it not a problem?*

**Reba:** *It's not a problem.*

**Irene:** *Why is it not a problem?*

**Reba:** *It's not.*

**Irene:** *No, you can't say it's not a problem.*

**Reba:** *It's not a problem for me, it's not.*

**Irene:** *Why is it not a problem for you?*

**Reba:** *It's not.*

**Irene:** *Mhm?*

**Reba:** *It's not.*

**Irene:** *You think it's ok to steal other people's food or to be constantly hungry?*

**Reba:** *Yah, am constantly hungry.*

#### **7.4.2.2 Skin picking**

According to Gourash and Foster (2005) the skin picking behaviour of PWS has a wide range of severity from patient to patient and sometimes in the same patient over time. Some patients have occasional minor skin picking while others maintain large open wounds. It has been related to boredom and anxiety. Reba started skin picking during the interview and he did throughout the interview until blood came out. He mentioned that it was not painful.

#### **7.4.2.3 Milestone developments**

Reba reported that his speech was delayed and the orthodontist had suggested a plate to force him to use his tongue in a different way.

#### **7.4.2.4 Medical problems**

I noticed that Reba had studs on his teeth, and I enquired about them. Irene informed me that she had taken Reba to an orthodontist for teeth alignment, but that he had removed the braces the same day, even though they had explained to him why he had to wear them. Reba reports that he removed his braces because they were painful, which is interesting because individuals with PWS are reported to have a high pain threshold, and Reba also agreed he was skin picking because it was not painful. Emerick and Vogt (2013) confirm that hypothalamic dysfunction has been implicated in many manifestations of PWS including the high pain threshold.

#### **7.4.2.5 Treatment received**

Reba mentioned that he goes to the doctor when he has flu and he has two other doctors who he consults with one for his teeth and another one for growth hormone therapy.

*“One is for my teeth, taking out my teeth, and then the other one is for, what you call it, gross hormone [sic].”*

#### **7.4.2.6 Schooling**

Reba mentioned that he is currently attending Raslouw Academy, and he is in grade five, but does not want to do matric at the school because the matric teachers shout at their learners.

*“I know, I know they shout at kids, they shout at matric kids, they shout.”*

#### **7.4.2.7 Food-related problems**

Reba admitted he had satiety problems but when he is at home he does not just take food, he actually asks for food.

*“Yah, am constantly hungry.”*

*“I ask.”*

However, at school Reba steals other children’s food and does not feel embarrassed or bad about it. When asked how he feels about it, he said:

*“Nothing.”*

Reba mentioned that he does not know why he cannot eat certain foods or why he cannot eat as much as he likes. He does not like eating healthy foods like fruits and vegetables; he prefers to eat braai meat.

*“Ha-ah I don’t, you didn’t tell me what it does, I just eat and she doesn’t tell me.”*

*“I don’t, I don’t eat it ... I like braai.”*

#### **7.4.2.8 Temper tantrums (outbursts)**

Reba mentioned that he sometimes has outbursts at school and the teachers call his mother but they don’t call her when he steals.

*“If I have tantrums they call you, that’s the secret, that’s the secret they don’t tell you. Only when it’s tantrums they call you.”*

#### **7.4.2.9 Weight control**

Reba was bought a bicycle for exercising, but he does not ride it because he fell when using it.

*“Mhmmhm, no, I am not. I don’t want to ride my bicycle. I won’t do it because I fell – mmhmm I am not going to.”*

#### **7.4.2.10 Siblings**

Reba confirmed he is looking forward to having a sibling and will help his mother take care of the baby.

*“Mmmhmm, take care.”*

#### **7.4.2.11 Hobbies**

Reba mentioned that he enjoys playing games on the phone and watching television.

*“I ask for a phone to play with.”*

He also enjoys playing with his friends in the park and going to Spur, to the shopping mall, and also to the movies.

*“Park.”*

*“The only thing its Spur, movies, going to the mall, and shopping – that’s the only thing that makes me happy.”*

#### **7.4.2.12 Social life**

Reba mentioned that he has friends at school and in the estate where they stay.

#### **7.4.2.13 Reba’s relationship with his father**

Reba was significantly impacted by the death of his father. He believed that without his father he was practically left alone. He reported missing him severely, and missed the activities they used to do together as a family. He spoke about his



father's death in a highly emotive manner. This indicated that the death of his father had a significant impact on his life.

According to Jarratt (1994) children have different reactions and responses to death and loss such as separation anxiety, impaired ability to make emotional attachments, anger, sadness, guilt, shame, depression, despair, problems with control issues, and diminished developmental energy.

*"But now am left alone, now am left alone ... I only have a mommy."*

#### **7.4.2.14 Routine and structure**

Reba emphasized that he does not like routine. He did not agree with his mother's idea of a routine. Part of his routine is to brush his shoes when he comes back from school, but he does not perceive it to be a routine and does not see the rationale behind polishing his shoes every day even though he only has one pair of black shoes.

*"Mmmm, that's not a routine ... Mmmhmmmm! No! I don't like routines, I don't want to do it anymore, I don't like routines. No! I don't. I am not happy with routines – mmmhmmmm NO! NO!"*

This is in line with the findings of Butler et al. (2006) that children with PWS do not easily agree to anything, are resistant to new ideas and experiences, and are more dependent than typically developing children.

#### **7.4.2.15 Future plans**

Reba reported that he wants to follow in his father's career path in the future – even though he did not have a clear idea of what that entailed.

*"Just be my dad's work [sic]."*

### **7.5 Collateral and additional information**

Potential behavioural problems present in most children with PWS require special education and support services (Goff, 2006). A psychologist's role is important during the schooling years. When the child is in junior high school, educators

frequently seek psychological advice. Psychologists are therefore faced with important issues when managing individuals with PWS. These include school placement, management of food-related behaviours, and problems like stubbornness and temper tantrums. The need for psychological advice seems to occur at predictable intervals as PWS individuals grow up. A request for a clinical re-evaluation or referral to a psychologist is generally precipitated by a behavioural crisis either at home or at school (Sulzbacher, 1988).

This was evident in this case; when I contacted Irene for a follow-up appointment she informed me that Reba had been suspended from school due to behavioural problems. She further mentioned that Reba was also suspended in 2015 due to the same behavioural problems, and was home schooled at the beginning of 2016. Irene had a scheduled meeting with the principal to discuss a way forward. The next session was then planned to take place after she had met with the principal and had discussed possible solutions for Reba. However, Irene contacted me and requested that I join her for the meeting with the school principal in order to get a comprehensive idea of Reba's behaviour in the school environment and to possibly help them to manage him better.

We met at Reba's school, but the principal was not able to meet with us due to personal matters she was attending to. Therefore, she asked the deputy principal, Mrs. Ellis, who also is in charge of discipline at the school, to meet with us. At the beginning of the meeting Mrs. Ellis provided the context, that, as the management of the school they never give up on a child because they cater for children who cannot cope in a mainstream school for different reasons. Some cannot read and/or write, some are slow readers, and some need remedial intervention. The school caters for children with different disabilities. Mrs. Ellis emphasized they are informed and educated about Reba's syndrome and understand it, and every time they employ a new teacher they explain it to them. This is because Reba tends to take advantage of the situation and returns to his old habits which they had already tried to manage.

Mrs. Ellis gave an overall description of the problems they experience with Reba. I have summarized these from the recording in a point format below. She mentioned the following problems:

- His behaviour varies from mild to severe and when it is severe it is to the point that it gets out of control.
- Reba will go to class in the morning and attend the first lesson and afterwards he will wander around the school yard and refuse to go back to class. They struggle to get him back to class as he becomes verbally aggressive and stubborn. Of late, he does not want to go to class anymore.
- When he is being disciplined he throws tantrums, shouts and then runs off.
- While wandering around when Reba notices that an office or class is not occupied he goes in and starts searching for money in order to buy snacks at the tuck shop.
- In the past Reba would steal money and go to buy things directly from the tuck shop. When they became aware of this they contacted Reba's parents, and, together, they agreed that the parents would give him a note so that he can go and buy from the tuck shop for the stated amount. In the beginning Reba's father would give him the note when he was going to school and sign it, but Reba started faking the notes. Then the parents asked the teacher to give him a note, but he again manipulated the system and he took advantage of the situation, and went to different teachers to get the note.
- Reba steals food from the kitchen especially when they have birthday snacks.
- Reba sits with the workers at lunch time when they are eating, so that they can give him food.
- Sometimes Reba goes to the staff room and makes coffee for himself, even though he knows that pupils are not allowed in the staff room.
- Reba was caught searching other learners' bags and when confronted he threw himself on the floor and refused to move.
- He also steals from the teachers. He stole R200 from a teacher and when he was confronted he admitted to taking the money.
- Reba goes into the finance office looking for money.
- He would also go into an unoccupied office and dial the different extensions and the phones will be left ringing off the hook.

Mrs. Ellis stated that they discipline pupils by giving them warning slips that they take home, and the parents sign these and after three warnings they have detention.

They then record everything and it goes to the discipline file of the pupil. The file has

to be updated by the teachers every day and brought back to her office by the end of the day but she never receives Reba's file. All their discipline measures do not work with Reba. It seems they use the same discipline techniques with Reba and other learners who do not have PWS, and it has been ineffective.

Irene confirmed the presence of these problems and added a few more behavioural problems which the deputy principal confirmed:

- Reba locks himself in the school bathroom.
- He throws orange peels at the acting deputy principal.
- He locked one of the teachers inside his office.
- Once Reba pulled his pants down and walked around naked in the school, while other pupils were watching him.

They both described Reba as intelligent, and always one step ahead and manipulative. They mentioned that his stealing methods seem to improve with the passage of time. The deputy principal added that Reba is not social at school, because nobody plays with him; he plays alone, and wanders around but he doesn't have friends.

*"He doesn't socialize. He does not interact with other children, [and] therefore the social aspect is not there even though he is amongst other children he is just causing disruptions."*

Reba's behaviour has seemed to get worse every year. In the first year (2013) the parents were called to school fewer than ten times, the following year it was weekly (2014), and in 2015 it was almost every other day. Victor went to fetch him three times in a week, because the teachers could not cope with him.

The deputy principal recommended home schooling for Reba highlighting that being in a normal school environment does not seem to be feasible for Reba. Transferring him to another school was also not an option for both Irene and the deputy principal. By the end of the interview, reading between the lines, it was clear they were expelling Reba from the school.

Reba's problems seem to fit the pattern for individuals with PWS: by school age, in addition to intellectual disabilities, social difficulties become more apparent (Whittington & Holland, 2010). They exhibit higher overall behavioural disturbances compared to individuals with an intellectual disability of unknown cause (Ho & Dimitropoulos, 2010).

I was taken aback by all the information I got from the deputy principal and the primary caregiver regarding Reba's behaviour at school. As I was listening, I started to wonder if the described behaviours could also be related to another physiological cause; I was considering the possibility of epilepsy, oppositional defiant disorder, secondary encopresis, and enuresis. My concerns were supported by the PWSUK (n.d.), that it is important to consider that certain aspects of PWS behaviour stem directly from physical disturbances in the brain.

## **7.6 Interventions**

- ***Client-centred therapy***

The client-centred approach was used with this family, as indicated earlier. This approach was founded by Carl Rogers and emphasizes three core conditions: empathy, unconditional positive regard, and congruence. They are not formulated as skills to be acquired, but rather as personal attitudes or attributes experienced by the therapist, and communicated to the client in order for therapy to be successful. According to Rogers, if therapists communicate these attitudes, the client will become less defensive and more open to themselves and their world – and will behave in prosocial and constructive ways (Corey, 2009).

The key characteristic of empathy is understanding another person's subjective reality as they experience it at any given moment. This requires an orientation toward the client's "frame of reference". Accurate empathic understanding implies that the therapist will sense a client's feelings as if they were their own – without becoming lost in those feelings. Unconditional positive regard refers to the experiencing and offering of a consistently accepting, non-judgemental and valuing attitude toward a client. The caring is non-possessive and is not contaminated by evaluation or judgement of the client's feelings, thoughts, and behaviour – as good or bad. Congruence implies that the therapist is real, genuine, integrated, and

authentic. The therapist openly expresses feelings, thoughts, reactions and attitudes that are present in the relationship with the client (Corey, 2009).

- ***Grief counselling***

According to Worden (2009) the loss of a significant other causes a broad range of grief reactions which are perceived as normal after such an experience. Most people can cope with these reactions and adapt to the loss. However, some experience high levels of distress. Grief counselling involves facilitation of uncomplicated or normal grief to a healthy adaptation within a reasonable time-frame. The overall goal of grief counselling is to assist the survivor to adapt to the loss of a loved one, and to be able to adjust to a new reality without the person concerned.

The aim of the interviews with Irene and Reba was to gather information on their experiences with PWS. However, in this process they presented with much sadness and distress about the death of their loved one. Irene was struggling to cope with the death of her husband and Reba was struggling to cope with the death of his father. This was evident, in that Irene presented with sadness most of the time, and could not fully focus on her daily activities especially taking care of Reba. Victor's death caused additional stress in her life, such as having to handle difficulties with Reba alone, and dealing with the pregnancy alone without her support system. Worden (2009) highlights that women who do not cope with bereavement tend to be young, with children, and with no close relatives nearby to help form a supportive network.

My hypothesis was therefore that Irene may not be able to give her full attention to and concentrate on Reba which is required when managing him. It is crucial for the primary caregiver to be mentally sound and emotionally contained in order to manage the individual effectively. This statement concurs with Whitman (1995) who states that the quality of life of the individual with PWS depends on the ability of caregivers to provide an environment which is structured enough for the individual, in order to minimize temptations, and that maintaining such a structure is labour intensive for parents and other caregivers. Moss (2009) also found that heavy demands are made on parents and caregivers of individuals with PWS, which usually lead to negative experiences and feelings. Whittington and Holland (2010)

added that families of people with PWS are subject to stress, and 70% of mothers have high levels of stress and need psychological counselling. Cassidy and Driscoll (2009) suggested that it is important to not only focus on the diagnosed individual, but also to provide necessary intervention to the primary caregiver, as they are confronted with multiple stressors.

Reba's behaviour at school and at home got worse following his father's death. He had frequent temper tantrums, was always talking about funerals, and was scared to sleep alone. The PWSUK (n.d.) mentioned that one of the factors that can worsen behaviour in individuals with PWS is bereavement. Therefore, I hypothesized that if Reba did not receive counselling then the obsessions with funerals would be intensified and tantrums may become frequent. This hypothesis is confirmed by White (1998) and Gourash and Forster (2015): individuals with PWS are stress sensitive, and therefore stress and anxiety are the source of most of their behavioural problems especially disruptive and aggressive behaviours, self-injury and tantrums. It was therefore important for me to be client-centered, ethically responsible, and to focus on the here and now, while addressing the presenting complaint. For this reason grief counselling was done.

Grief counselling was done with Irene during the first session, and with Reba during the second session. During the fourth session they were seen together as Reba had requested to see me stating that I was the only person he could talk to about his father. Worden (2009) found that the most significant losses occur within the context of a family unit, and it is important to consider the impact of a death on the entire family system. Most families exist in some type of homeostatic balance, and the loss of a significant person in the family group can unbalance this homeostasis and cause the family to feel pain and to seek help. This was particularly true for this family, and therefore it was important for me to have a session with both of them.

Irene and Reba were allowed the space to talk about the circumstances around the death. They shed their memories and connected over these – and for the first time the conversation was flowing and there were no arguments. Reba had a glow and smile on his face as he spoke about his father, and this rubbed off onto the mother. The aim of this was to help them actualize the loss. This is supported by Worden

(2009) who stated that one of the best ways to help the person actualize the loss is to help them talk about it, and also the circumstances surrounding the death. Many people need to go over and over the death in their minds, reviewing the events of the loss, before they come to the full awareness that it had happened. They were given empathy through this process, which mobilized them to talk about their feelings about the loss, and which brought emotional relief to both of them. This has been acknowledged as being the part of the grief counselling that it is important to help the person to identify and experience feelings (Worden, 2009). Larson (2013) supports this by reporting that a person-centred counselling approach (described earlier) with an emphasis on a deeply empathetic and caring therapeutic relationship precisely fulfils this fundamental criterion for effective grief counselling.

Reba mentioned that he has now created a special place for his father in his heart, and that he felt better compared to the previous session. This suggested to me that the process automatically facilitated emotional relocation of the deceased. According to Worden (2009) by facilitating emotional relocation, the counsellor can help the survivor find a new place in their life for the lost loved one – a place that will allow the survivor to move forward with their life and to form new relationships. Irene and Reba were both emotionally contained and were grateful for this process. Their grief reaction and process were then normalized for them.

- ***Behaviour therapy***

The second, third and fourth sessions also incorporated behaviour therapy. According to Corey (2009) behaviour therapy practitioners focus on observable behaviour, current determinants of behaviour, learning experiences that promote behaviour change, and tailoring treatment strategies, assessment, and evaluation. The aim was to define the problem, identify what needed to be changed, assess the effectiveness or ineffectiveness of the current intervention, and devise intervention strategies. Grant and Evans (1994) advised that when facing a behavioural problem, one of the first things to do is to identify what needed to be changed. They further emphasized that it is important for the behaviours to be clearly defined.

### ***Second session***

- ***Identifying the problem and what needs to be changed***



With the second session, and after the interview with Reba, I observed him having an outburst just before I left. The tantrum was directed at his nanny. I wondered if the tantrum was not perpetuated by the fact that during the interview he had just spoken about his father and was emotional. He kept talking, and while tears were streaming down his cheeks, he kept repeating: *“I am so upset right now.”*

- ***Social reinforcers and Pivot tool***

Grant and Evans (1994) state that social reinforcers involve the actions of other people – for example, response-dependent praise, smiles, kisses, and applause that strengthens desired behaviours. Social reinforcers are highly effective for most people, and are important determinants of our everyday interactions.

I thought of a way to calm Reba down by distracting him and asked him to get me some toilet paper. He was still able to cooperate and comply with the request. I then acknowledged him, complimented him on his positive attributes, and this seemed to calm him down. As a result, the outburst didn't last long. Using social reinforcers seemed to be effective in managing this episode. The maintenance of positive social contacts with the person, praise, and stressing the PWS person's positive aspects, are likely to have a positive effect on them (PWSUK, n.d.). This is supported by Whitman and Jackson (2006) who described the praise and ignore (or pivot tool) which illustrates that when inappropriate behaviour occurs, the strategy is to pivot attention away from the problem behaviour to another task or the appropriate behaviour of another individual, to wait patiently for the targeted individual to engage in some appropriate behaviour, and then pivot attention and praise back to the individual.

### ***Third and fourth sessions***

- ***Identifying the problem and what needs to be changed***

The following were identified as problems:

- a) The temper tantrums were identified to be a problem. They are:
  - frequently observed at school;
  - mostly directed at Boitumelo;

- also directed at certain people, but not at everyone; we identified the people that were mostly the “victims” of these outbursts, and found them to be Boitumelo, Reba’s aunt, the older uncle, and his cousin. Irene described them as inoffensive, gentle and sweet;
- frequently around bath time especially with Boitumelo and also happen when Reba is being told what to do.

b) The nanny (Boitumelo) who is inconsistent

- Irene reported that when they employed the nanny they stressed that she must not try and be friends with Reba. Reba is naturally friendly and it is easy to establish good rapport with him but he needs a disciplinarian and someone who will adhere to the schedule. However, in this regard, the nanny did not comply.
- Boitumelo often rewards Reba with food, and then the next day she tries to be strict with him. She sometimes changes his diet schedule because she feels sorry for him. Irene emphasized that with Reba, once you have presented with inconsistency and lack of structure he will always perceive you as presenting with such, even if you try to change and adhere to the schedule and become consistent. His perception of you has, however, been distorted and will not change. Reba will refuse to obey you and follow your rules. He will just say: *“Ah, that’s ausie [Boitumelo] I know; she is not going to do it.”* Then he will wait for her to fail and will say: *“I told you that she won’t do that.”* When she tries, he will tell her: *“you just doing this for today.”*

Whitman (1995) emphasized that consequences must be enforced without fail. Any change leads to a toppling of the whole structure as the person with PWS is strongly attuned to other peoples’ vulnerabilities and can manipulate them almost at will. The PWSUK (n.d.) states that the individual may “act up” in one environment where their behaviour is “rewarded” by others “giving in” but will not exhibit the behaviour in an environment where others take a firmer stance. This was observed with Reba.

c) Reba’s behaviour:

- Reba does not want routine and when there is one in place, he does not adhere to it.
- Reba tends to be argumentative.
- Reba reported that he sometimes steals food.

- ***Rule-governed behaviour***

Some of the interventions below, where instructions are used, highlight the effectiveness of rule-governed behaviour. Rule-governed behaviour is often planned, calculated, logical, and the person whose behaviour is rule-governed knows the rules (Grant & Evans, 1994). According to Grant and Evans (1994) changing behaviour with instructions is rule-governed behaviour. In full form, rule-governed behaviour is the behaviour of following the rule.

There seems to be a correlation between consistency and tantrums. Consistency has proven to be effective with Reba thus far, as he does not throw tantrums around Irene and his uncle. The reason for this is because:

- Irene has made the rule that when Reba has an outburst, he must go to his room and calm down.
- She ignores him when he has an outburst; McCandless et al. (2011) state that behavioural management that focuses on rewarding desired behaviours and ignoring, when possible, undesirable behaviours seems to be most effective.
- Irene does not negotiate with Reba about bath time, and instead if he refuses to comply she threatens him with a belt: he responds better to strict rules and firmness.
- She described herself as structured and organized.
- Irene adheres to the schedule, even if the schedule disadvantages her.
- Reba's uncle is also very strict and firm with him.

Irene believes that it is important to: *“apply tough love and be consistent.”* She further elaborated that it is very important that you must be consistent with Reba because if there is no consistency then they experience problems. This has also been observed at school with teachers who do not implement discipline consistently, as Reba tends to take advantage of the situation.

During our discussion, Irene realized that there is a high probability that the nanny lacks consistency. The plan was then to talk to the nanny about the importance of consistency and firmness in managing Reba without fail in order to restore order to their relationship. It was crucial that the nanny also adhere to the scheduled dietary plan Irene designs.

- ***Anticipatory planning***

The other method which Irene has been using and which has been effective, is that she will tell Reba how much money she has in her possession to make him aware that she knows how much there is so that he won't steal the money. She also informs him in advance if there are going to be changes in his diet schedule for the day. This is in line with the assertion that good management means always anticipating tomorrow, next week, new situations, changed plans, and the like. Clearly delineated authority figures and anticipated rewards and consequences, provide the structure for behaviour with clear limits and without ambiguity (Whitman, 1995).

- ***Social reinforcers***

At the beginning of the fourth session, Irene gave feedback on Reba's tantrums, which have been better since the grief counselling which took place in the second session; she now described Reba as well behaved. I made use of a social reinforcer when I commended him for his efforts. This was done through label praise which was very encouraging for him. Label praise (also known as descriptive praise), is praise that specifies the behaviour or features of behaviour which are desirable (Grant & Evans, 1994).

- ***Positive reinforcement***

Positive reinforcement involves the addition of something of value to the individual (such as praise, attention, money or food) as a consequence of certain behaviour. When the goal of a programme is to decrease or eliminate undesirable behaviours, positive reinforcement is often used to increase the frequency of more desirable behaviours which then replace undesirable behaviours (Corey, 2009).

We discussed a reward system when Reba adheres to his scheduled diet and daily routine, and he suggested that he could be rewarded by going to the movies. We negotiated the snacks he could have at the movies, which would not exceed his calorie intake for the day. I gave him an opportunity to come up with a trial period, and he suggested a month. We also decided that I would check-in with him to see if he was still adhering to the programme and keeping his promise. Positive reinforcers are generally not difficult to identify and reward systems that use small, short-term goals that progress to larger goals are quite effective (McCandless et al., 2011).

As a family they have previously tried a reward system, but they did not use food as a reward. They rather used activities outside the home. They are weary of Reba doing well, just to get the food which will cause problems for him. This is in line with McCandless et al. (2011) who advised that parents should be counselled that offering food as a reward or withholding food as a punishment, is almost always counter-productive and should be avoided.

A follow-up was done two weeks later to determine if Reba was complying with his diet, and Irene reported that he had been doing very well as he has been adhering to his diet plan, and Reba confirmed that he was keeping the promise he made. His efforts were acknowledged. This intervention seems to be effective in the short-term, but Reba tends to go back to his usual habits after some time. Although the family had also been implementing a reward system, there has been no significant improvement because food-related problems persist with Reba. A long-term intervention and solution is needed.

- ***Patient participation***

Patient participation means involvement of the patient in decision-making or expressing opinions about different treatment methods which includes sharing information, feelings, and accepting health-team instructions. With enhanced patient participation and considering patients as equal partners in healthcare decision-making, patients are encouraged to actively participate in their own treatment process, and thus follow their own treatment plan (Vahdat, Hamzehgardeshi, & Hamzehgardeshi, 2014).

As part of the intervention regarding Reba's resistance to adhering to routine, in the second interview I considered that if Reba were involved in structuring the routine, it might assist him to feel he is still in control and to feel empowered in the process but this was ineffective as he stubbornly insisted that he did not want to have a routine, and could not suggest any alternatives. It was clear that Reba does not respond positively to being told what to do or being asked to engage in a certain way.

During the fourth session after rapport was established, I suggested to Reba again that he should assist his mother to structure the routine. Although he did not want to be involved in planning the structure, he did agree that Irene could decide on the structured routine, and that he would adhere to it. However, he still took control and gave his mother instructions. It was his decision to make and he was not told what to do. A follow-up was done two weeks later, and Irene reported that Reba was calmer and usually complies with his routine compared to previously, even though he occasionally gets stubborn.

- ***Feedback intervention***

Feedback intervention providing people with some information regarding their task performance is one of the mostly widely applied psychological strategies (Kluger & Denisi, 1998). According to Hattie and Timperley (2007) feedback is conceptualized as information provided by an agent regarding one aspect of one's performance or understanding. It typically occurs after instruction that seeks to provide knowledge and skills or to develop particular attitudes.

The aim of giving feedback was to make Irene aware of her contribution in maintaining the undesired behaviour. Feedback was given based on my observations that Reba is stubborn and likes arguing about issues and that Irene tends to "feed" the argumentative style by arguing back. Feedback on her contribution was given with the aim of minimizing the arguments that take place between them.

Contrarily Irene explained that if you do not argue with Reba, he will feel he has won and that what he is saying is right because he wants to win every argument. It was also evident that after a lengthy argument with Irene, Reba would, however, finally

give in or agree. This also contradicts the views of the PWSUK (n.d.) that one should never argue with an individual with PWS, because you will not win an argument, and that you need to take a break especially if the person is becoming vocal and abusive.

- ***Psycho-education***

According to Lukens and McFarlane (2004) psycho-education is among the most effective of evidence-based practices that have emerged in clinical trials and community settings. It is a professionally delivered treatment modality that integrates and synergizes psychotherapeutic and educational interventions. Psycho-education reflects a more holistic and competent-based approach, and stresses health, collaboration, coping, and empowerment. A psycho-education model therefore views the role of the psychological practitioner, not in terms of abnormality, diagnosis, prescription, therapy, and cure but rather in terms of client dissatisfaction, goal-setting, skills teaching, satisfaction or goal achievement (Authier, 1977).

During the fourth session, Reba described himself as: “not well behaved” when he steals food. He further said that he attended a friend’s birthday party and they went ten-pin bowling, and had pizza, happy meals, cake, and snacks. Irene was surprised at this revelation, as she was told a different story. He went with the friend’s parents who are aware of Reba’s condition but they still allowed Reba to eat that much. I then acknowledged the challenge he is experiencing regarding self-control when it comes to food, and he accepted the empathy. I used this opportunity to psycho-educate him on the consequences of over-eating.

Two months later Irene gave feedback that Reba was adapting well to having a sibling and his behavioural problems especially the tantrums had improved but they were still experiencing challenges with controlling his food-related problems. He was now enrolled at Montessori; his previous school and he is also attending sessions with their school psychologist.

## **7.7 Conclusion**

This chapter demonstrated that having a child diagnosed with PWS had a significant negative impact on the parents. They struggled with accepting the diagnosis and taking care of their child with PWS disrupted Irene’s studies, it was time consuming,

it affected Victor's work schedule, and it was emotionally overwhelming for them. They were pessimistic about the future.

The chapter also relates the tragic story of Irene whose husband died recently; she was expecting another child and was now faced with the sole responsibility of taking care of her son Reba who has PWS. The parents' intention to keep Reba's condition quiet is in tatters as it prevented Irene from getting adequate support after her husband's death. It is clear that Irene is in dire need of social support. I intend to keep in contact with her in order to be of some assistance in this regard.

Reba presents with severe behaviour problems. Some of the behaviour problems such as food-related problems; satiety; temper tantrums; manipulative tendencies; lying; obsessive-compulsive behaviours; argumentativeness and stealing are in line with what is documented in the literature. What I found different with Reba is that he also had elimination disorders – encopresis and enuresis. Reba's behaviour seemed to progressively worsen every year and subsequently he was out of control in his new school and has been expelled. At home, Reba's nanny cannot contain him and sticks to a regulated feeding regime. He was treated with Concerta by a neurologist to try and manage the behaviour problems.

Another challenge for this family was the fact that because Irene gave birth in a private hospital they were not able to get GHT and their medical aid excluded it. They had to finance it for a short period which proved to be very expensive, subsequently they had to stop it and Reba's growth was compromised. This suggests that it is crucial for medical aids in South Africa to consider classifying GHT as a chronic medication for individuals with PWS.

I did my best to assist this family. In so doing, my role of researcher was to some extent overshadowed by my attempts to provide therapy. In this respect I made numerous contacts with the family, got collateral information from the school and applied grief counselling and behaviour therapy. Following the grief counselling, Irene was able to focus on Reba and Reba presented with decreased temper tantrums and showed a willingness to adhere to his dietary schedule. A combination of social reinforcers; rule-governed behaviour; positive reinforcement; anticipatory



planning; patient participation and psycho-education proved effective in managing some of the behavioural problems. Changing schools and attending sessions with the school psychologist also seem to have contributed to improved changes in Reba's behaviour particularly within the school environment.

## **CHAPTER 8**

### **STEPHANIE'S STORY: "THAT'S ALL I WANT"**

#### **8.1 Introduction**

In this chapter I will describe the lived experiences of Anna, the primary caregiver, and her daughter Stephanie, who has been diagnosed with PWS. First, I will illustrate how PWS can be easily misdiagnosed, how the parents had to adapt to having a "sick" child, and the challenges they experienced with the nurses. I will also highlight the impact that the diagnosis had on the parents, and especially on Anna. I will also describe how PWS affected Stephanie's physical health. Also discussed is how religion can prevent an individual from receiving optimal treatment.

Secondly, I will demonstrate that caring for an individual with PWS is emotionally exhausting and time-consuming for the primary caregiver. Thirdly, I will illustrate the difficulties experienced in managing the behaviour problems associated with PWS, and how these problems also manifest in the school environment. To conclude, I will give details on different interventions implemented, and also their effectiveness.

The sessions with Stephanie and Anna were conducted in my office at the Dr. George Mukhari Academic Hospital. One session was conducted at the school. The first session was held towards the end of October 2011. They then attended monthly sessions until March 2012. From May 2012 they attended bi-monthly sessions until October 2013. They came back to me in April and June 2014 for follow-ups, where they gave feedback on the behaviour and interventions they were implementing. The last two sessions they attended were in August 2015 and February 2017.

The information from this family was gathered at different times even before I started with the present research. Although some of the information was documented in the form of process and progress notes, most of the sessions were voice recorded, which made it possible to transcribe and utilise them for the research purposes of the present study. The quotes from the voice recording were translated from Setswana into English.

This *modus operandi* is supported by Wolfson and Sampson (1976) who state that process notes may be used effectively for some psychotherapy research, particularly those that deal with thematic shifts that occur over a period of time.

## **8.2 Family background**

I first met this family in 2011. At the time, it comprised the father, the mother, and three children. The mother and father separated in 2013 and divorced in 2015. The family resides in a small, one-room RDP house with an outside shack, in Mothutlung, a township in North West Province. The area is mostly populated by black Africans of lower and working class. Their family income is R3800, and they are currently struggling financially. They are Jehovah's Witnesses and attend their church twice a week, on Wednesday and Sunday. As they do not own a car, they have to walk to school, and they use taxis when they travel longer distances. Their home language is Setswana. The family is affiliated with the PWSSA.

## **8.3 The context**

This family was referred from the Paediatrics Outpatient Department by Dr. Duncan. The referral stated that Stephanie was presenting with stubbornness, temper tantrums and disobedience, and they required psychological intervention for these behaviours. This is the family that introduced me to PWS for the first time.

Anna is a 48-year-old, African divorced female. She passed grade 12, but could not further her studies as she got married at the age of 18. She has been working as a hawker since 2013 selling cosmetic products to earn extra income. In addition to this, in 2015 Anna was employed as a cook at Stephanie's school, but her contract there ended in March 2017. She is friendly and warm. She gave me reading material on PWS and DVDs to watch which helped me gain a better understanding of PWS.

Stephanie is an 18-year-old, African, single female. She is the first-born child in the family. She was 13 years old when I first met her, but she appeared younger than her stated age. Stephanie has a short stature and is obese, and throughout the time I saw her, her weight seemed to increase. She has observable dysmorphic features, gait difficulties, evidence of skin picking around her nails and fingers, and on her chin, she wears a scoliosis brace.

Stephanie is currently enrolled at Neo Mathabe, a special school in Mothutlung in North West Province. She is a recipient of a disability grant. Stephanie presented as friendly, warm, spontaneous, and talkative. These positive characteristics concur with IPWSO (2013) which mentions that people with PWS have many positive characteristics, are known to be friendly, sociable, kind and caring, and many have a wonderful sense of humour.

During the first session Stephanie was pre-occupied with toys as she entered my office, and she took some of the toys and put them in her bag. When she was tired of playing with the toys she became restless. As I was doing an intake interview with Anna, she kept interrupting and arguing with her. In an interview with Stephanie, I noticed that she could not comprehend information, was not aware of the impact of her behaviour on others, her cognitive functioning seemed to be impaired, and her thinking was concrete.

#### **8.4 Lived experiences**

The lived experiences of Anna and Stephanie were explored during sessions held with them. Certain themes were identified which are discussed in the following sections.

##### **8.4.1 Themes identified from interviews with Anna**

During the interviews and sessions with Anna, the following themes became evident:

###### **8.4.1.1 Pre- and post-delivery**

Anna reported that in the beginning of her pregnancy the foetal movements were normal. However, as her pregnancy progressed the movements decreased significantly until they almost ceased. She informed the nurse at the clinic, but they could not assist her. They told her to wait until delivery. Because it was her first pregnancy, she was unsure if it was normal. This experience concurs with the research findings of Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) that in individuals with PWS, hypotonia usually manifests as decreased foetal movement.

*“In the beginning she was playing and kicking. After some time, she would kick once a day and nothing more.”*

Anna’s pregnancy was full-term and she had normal vaginal delivery. This experience is atypical. According to Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) in individuals with PWS, hypotonia usually manifests as abnormal foetal position at delivery, and there may be an increased need for assisted delivery or a caesarean section.

Anna mentioned that Stephanie did not cry after delivery, she did not have energy to eat, and her hands and legs were floppy. She described the floppiness as being like chewing gum (Chappies). Stephanie was sleeping and did not feed the whole night and most of the day. When she told the nurses, they tried to wake the baby by tugging the feet, but this did not work. Anna only got assistance when her husband came for a visit and confronted the nurses. It was only then that they called the doctor. The doctor found the sugar levels fluctuating between high and low. Stephanie was then monitored every 30 minutes. A feeding tube was then inserted into her nose. In infancy hypotonia is also characterised by decreased movement and lethargy, with decreased spontaneous arousal and weak crying (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

*“They inserted a tube in the nose so that she could feed.”*

*“She was a Chappies.”*

*“She didn’t cry.”*

Anna gave birth in Jubilee Hospital in Gauteng province. After a week they were transferred to Kalafong Hospital, where they were admitted for three weeks.

#### **8.4.1.2 Feeding problems**

Difficulty with sucking is one of the commonest symptoms with new-borns having PWS. Stephanie was fed through a tube for one month. According to Cassidy and Driscoll (2009), Ho and Dimitropoulos (2010) and Cassidy et al. (2012) part of the characteristics of hypotonia are poor reflexes, including poor sucking, which leads to early feeding difficulties and poor weight gain.

#### 8.4.1.3 Diagnosis

In Kalafong Hospital Dr. Duncan conducted blood tests and Stephanie was initially diagnosed with glutaric aciduria type II.

*“Dr. Duncan was continuously doing blood tests until they diagnosed her with glutaric aciduria type II.”*

She was told that there were two children in Pretoria that had been diagnosed with glutaric aciduria type II. During every consultation she asked for feedback on the progress of the other two children. They could not tell her if Stephanie would be able to walk as children with glutaric aciduria type II cannot walk.

*“Will she walk? What’s going to happen to her? And they said they didn’t know.... I was always trying to get information ... it was stressful.”*

Anna reported that it was difficult for them as parents but they had to eventually accept their child’s condition.

*“When you get a sick child, you just have to accept what else can you do? Her dad cried until he was ok ... he was hurting badly.... He eventually accepted.”*

In 2005 Stephanie was seven years old and had been walking for five years. She was doing a lot better compared to the other two children. She was, however, eating a lot. Anna then requested that she stop giving the child multivitamins. She further requested that Stephanie be tested again. They conducted tests and Stephanie tested negative for glutaric aciduria type II. She was then tested for PWS, and tested positive. This is in line with the research of Caldwell and Taylor (1988) who gathered information from 12 families, and found that at the time of birth a diagnosis of PWS was extremely rare. Until the time that infants begin to demonstrate observable characteristics of the disorder, individuals are often misdiagnosed or not diagnosed at all. However, Whittington and Holland (2010) reported that compared to 20 years previously, there was a generation of children who had been diagnosed with PWS within days or weeks after birth. Bar et al. (2017) found that the mean age at diagnosis was 18 days. Stephanie was born in 1998, but was only diagnosed in 2005.

Anna reported that she never received any counselling and education relating to her daughter's anomalies. She had to find a way to cope. This agrees with Whitman's (2006) research findings that parents of children with PWS got less support from professionals compared with parents of similarly aged children with cognitive deficits resulting from other causes. This family also did not receive adequate support from healthcare professionals.

*"They do not take you for counselling; you counsel yourself."  
"I counselled myself."*

#### **8.4.1.4 Milestone developments**

Stephanie's developmental milestones were delayed. Anna could not recall all the information, but remembers that the baby was moving on her buttocks, she did not crawl. Stephanie started walking at about the age of two. Her speech was delayed. This finding is supported by Cassidy and Driscoll (2009) who said there are delayed milestones, including gross motor and language delays, in individuals with PWS. Early milestones are reached, on average, at double the normal age. For example, sitting occurs at 12 months, walking at 24 months, and speaking words at 2 years. Stephanie only started menstruating at the age of 17.

#### **8.4.1.5 Treatment received**

Following the glutaric aciduria type II diagnosis, Stephanie was prescribed L-Carnitine. They had regular follow-ups at the hospital. They consulted with an occupational therapist who trained Stephanie and taught them exercises to improve her floppiness and to help her with walking. They were also given neck exercises to strengthen the neck. They also attended a speech therapist, because Stephanie's speech was delayed. This concurs with Cassidy and Driscoll's (2009) suggestion that children with PWS should receive early intervention (including physical, occupational, and speech therapies) and also individualized, appropriate education. They received these interventions despite the wrong diagnosis. They continued to consult with the endocrine clinic, the genetics clinic, dermatology department, and the spinal unit.

Recently, Stephanie has been presenting with behavioural problems. As a result, Anna informed the dermatologist and amitriptyline was prescribed for her daughter. This is a tricyclic antidepressant (Sadock & Sadock, 2003). Dr. Duncan suggested that Stephanie take the medication for a week, and if there is no improvement they should inform her. Anna wanted something to calm Stephanie down – but the medication did not seem to be working.

#### **8.4.1.6 Sleep difficulties**

During her early and late childhood, Stephanie slept excessively. Although she slept during the day and at night she still appeared to be tired when waking up in the morning. Whittington and Holland (2010) stated that some children with PWS may have sleep disorders, including disruptions of the normal sleep cycle and excessive daytime sleepiness. Recently, Anna mentioned that Stephanie only sleeps at night and then she sleeps throughout the night. Stephanie only wakes up when she needs to go to the toilet.

#### **8.4.1.7 Medical problems**

Anna reported that Stephanie had pneumonia at about the age of three to four. She was also diagnosed with chronic sinusitis. She was once admitted to hospital for an abscess, following a skin picking incident. In 2013 the doctors confirmed that Stephanie had scoliosis. She was then given a scoliosis brace to wear every day, to try and prevent her back from deteriorating further but it is not very useful. Stephanie needs to undergo an operation to prevent further curvature of her spine.

Goelz (2006) reported that individuals with PWS are more at risk of neuromuscular scoliosis presumably as a result of low muscle tone. Approximately 62-68% of the PWS population have scoliosis, with a structural change of at least 10 degrees. The treatment for neuromuscular scoliosis ranges from careful monitoring, to bracing, through to surgical stabilization.

Anna faces a moral dilemma as to whether she should allow the operation as this might involve a blood transfusion which was prohibited by her church. If there is no other option it means that Stephanie will never undergo the required operation.



*“In our religion we do not do blood transfusion.”*

*“She will stay like this.”*

#### **8.4.1.8 Decision to have another child**

Anna mentioned that she did not plan to have another child after giving birth to Stephanie. Both her other two pregnancies were unplanned.

*“It was not my aim to have the second and the third. It was a mistake ... They were all mistakes.”*

#### **8.4.1.9 Schooling**

Stephanie never went to a crèche as she was enrolled in Neo Mathabe special school from the beginning. Eventually, however, Anna was not satisfied with the standard of education at Neo Mathabe. Therefore, she placed Stephanie in a Pretoria school. Unfortunately, she was expelled from this school after only six weeks, because she took medication from the sick room, locked herself in the toilet, opened the medication and refused to open the door. The principal at the school felt they could not give special attention to one child as their motto was: “majority rule, not minority rule”. Anna was thus compelled to take Stephanie back to Neo Mathabe, where she has remained until the present time.

Stephanie still cannot write her name and does not cooperate in class. This seems to fit the pattern, in that mental retardation is an integral part of PWS (Whitman, 1995) and cognitive disability is evident when PWS children start school. Most people with PWS have multiple, severe learning disabilities, and also poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010).

#### **8.4.1.10 Food-related problems**

In 2011, Anna reported that Stephanie asked for food and money from people. When she asked people for food, they got annoyed. Some gave her food whereas others refused. Stephanie stole food from the shack because Anna sometimes forgot to lock the shack which was where she stored the food. She suspected that Stephanie

also stole food from the school kitchen. Stephanie had temper tantrums whenever she was denied food. Anna used snacks to reward good behaviour.

During the 2017 interview, Anna mentioned there was a slight improvement in Stephanie's food-stealing behaviour in the home environment compared to 2011. However, if they forget to take food items back to the shack Stephanie eats what has been left over, even if it is burnt, stiff pap (porridge). When Stephanie has had access to food, she hides it in her brace or in her underwear.

*“She will take four slices of bread and shove them in her underwear, because she [then] knows we cannot eat them again.”*

Cassidy and Driscoll (2009) and IPWSO (2013) caution that lack of satiety should be acknowledged at school, work, and home and all caretakers and supervisors need to understand this. Consistent limit setting and close supervision are necessary at all ages. This is what Anna tries to implement most of the time.

Anna reported that when she (Anna) is not paying attention Stephanie sometimes takes advantage and asks people for money or food. When Anna intervenes, and tells the person not to give Stephanie food or money, she is sometimes met with resistance which makes it difficult for her to pre-empt the behaviour or try to be in control all the time. According to Bos, Pryor, Reeder, & Stutterheim (2013) this kind of response from people could be associated with the public stigma which presents people's social and psychological reactions to someone they perceive to have a stigmatized condition. The person believes that the stigmatized individual cannot be held personally responsible for their condition and this leads to feelings of sympathy for the stigmatized individual, and a need to provide help (Bos et al., 2013).

*“Sometimes when I tell people not to give her anything when she asks, they will respond by saying: ‘I don't want God to punish me. I will give her’”.*

Anna is worried about Stephanie as she is currently overweight. Stephanie sometimes refuses to adhere to the diet prescribed by the dietician. For example, she will not drink tea if she cannot taste the sugar in it.

*“When you tell her, she can’t have more than two teaspoons of sugar, she will bring the tea to you and say: ‘you drink it’ – and she provokes you.”*

#### **8.4.1.11 Weight control**

Anna reported that she tries to control Stephanie’s weight by following the diet prescribed by the dietician, and they all eat the same food. This is in line with IPWSO (2013) that it is imperative to begin good meal management and education at an early age. This includes sticking to a strict schedule for meals and snacks, and also limiting portion sizes.

#### **8.4.1.12 Behaviour problems**

In 2011, Anna reported that she was struggling to take care of Stephanie as she needs to be monitored all the time. She occasionally defecated at various places in the school premises, stole money, lied a lot, and would lock herself in the toilet. When they went to the mall Stephanie would disappear and ask people for money or ask them to buy her things. She did the opposite of what she was told to do. Stephanie was very stubborn and swore a lot. When she started doing something, she did not stop. Stephanie did not learn when she was taught through demonstration. She also took very long to bath.

During the 2017 interview, Anna spoke about Stephanie’s behaviour in an overwhelmed manner. She described Stephanie as naughty, and reported that she does not listen when she talks to her even when she is trying to guide her on what is appropriate or inappropriate behaviour. Stephanie interrupts her when she has conversations with people. She also took medication they got from the doctor, and drank half of each of the bottles and then added water to it. Stephanie drinks any medication she comes across and this may be dangerous for her.

*“When you tell her to stop – she doesn’t.”*

*“She is a real stressor; I find myself really annoyed.”*

*“Am not expecting her to listen 100%.”*

*“I feel like I am going crazy sometimes. She can make you lose focus of important things; it is too much, I am not coping.”*

At times, Stephanie takes her siblings' stationery and hides it and then she cannot recall where she has hidden it or takes it to school and gives it to other learners. As a result, her siblings get into trouble at school.

*“You see now it is a problem, because she loses other children's school books and stationery.”*

Anna also mentioned that Stephanie cannot be left alone in the house, because she breaks items in the house. For example, Stephanie broke a kettle while trying to make tea.

*“She breaks a lot of things.”*

Stephanie does not listen to reason, and she is very argumentative at school and at home. She continues to swear a lot: she swears at everyone, she swears at Anna and the siblings, she even swears at them in public when people are watching. She beat the neighbour's chicken to death, which had been strutting in their yard. Subsequently, Anna often has to take responsibility for Stephanie's behaviour, and apologizes to the neighbours, teachers, and even strangers.

*“As she was beating the chicken with a stick she kept asking: ‘what are you doing here’ – until it died.”*

Butler et al. (2006), Ho and Dimitropoulos (2010), and McCandless et al. (2011) established that individuals with PWS present with stubbornness, resistance to change and a tendency to be very argumentative, to lie, steal, and be aggressive. They do not easily agree to anything, are resistant to new ideas and experiences, and are more dependent than typically developing children. These characteristics are common during the childhood years and continue into adolescence and adulthood. Stephanie displays all these characteristics.

#### **8.4.1.13 Impact of PWS on Anna**

Anna stated that taking care of Stephanie is overwhelming for her. She thinks that she could possibly be depressed again. She mentioned that she was previously

treated by a psychiatrist for depression and anxiety because she was not able to cope but she defaulted on her treatment.

*"It feels like there are whistles in my head, I think I have depression ... I do not sleep well."*

*"I feel like I am going crazy."*

She reports that constantly taking care of Stephanie hinders her from getting a full-time job. She wishes she could find a home where Stephanie could be placed.

*"My life is on a standstill. Someone was even suggesting that I should also get a disability grant because I am not able to have a full-time job; she is my job, I am struggling."*

*"I wish there was a home where I could take her."*

Anna mentioned that Stephanie needs to be monitored all the time.

*"She needs to be monitored 24/7 – you need to follow her around."*

*"By the time you go to bed, you don't want anything, you are tired."*

Anna's experiences are supported by Moss (2009) who found that heavy demands are made on the parents and caregivers of individuals with PWS which usually leads to negative experiences and feelings. Whitman (2006) also highlighted that harried schedules, work overload, financial difficulties, and other external precipitants of stress may become overwhelming.

#### **8.4.1.14 Siblings**

Stephanie has two siblings, Lucky the second born, and Tinny the last born. In 2012, Anna reported that taking care of Stephanie was extremely difficult for her, because she also had two other children to take care of. Lucky had started stealing money and it also became difficult for her to deny the other children sweets and snacks because Stephanie could not have them. Anna was struggling to discipline all her children. I conducted a family intervention session with Anna and the children where psycho-education was done to assist the siblings to understand PWS and Stephanie's behaviour. Parental guidance was also done to assist Anna with

effective ways to manage and discipline the children. During the follow-up sessions Anna reported a great improvement.

In 2017, Anna reported that Stephanie and Lucky tend to fight a lot because Stephanie swears at him. Despite the conflict, the siblings are keen to assist in taking care of Stephanie when Anna is not around, which also places additional responsibility on them. This concurs with Whitman's (2006) finding that siblings of individuals with PWS are pressured into parenting roles or given additional responsibilities. This tends to be too much for Stephanie's siblings, especially when they want to go and play. Therefore, in order to cope, they lock Stephanie in the house.

*"Lucky ends up beating her."*

*"I leave her with Tinny and Lucky, [and] they take care of her."*

*"When Lucky wants to go and play he locks her in the house."*

The experiences of this family and the interventions provided concur with Mazaheri et al.'s (2012) findings that families, mothers and siblings of children with PWS reported difficulties in family functioning, communication problems, and an increased number of conflicts. The study reaffirmed that PWS affects the entire family system. Mothers and siblings would benefit from psychosocial support due to the multiple challenges of living, with and caring for, a child or young adult with PWS. The authors suggested that interventions should focus on providing psychosocial support to each member of the family. This should encompass all aspects of life, such as the emotional, physical, communication issues, family dynamics, and personal perceptions of disease. The family dynamics and their personal perceptions of the disease were both addressed with this family.

#### **8.4.1.15 Social life**

When Stephanie is at home they do not allow her to play outside with other children, because when she is allowed to do this, she asks people for money or she wanders around.

#### **8.4.1.16 Adaptive functioning**

Anna alluded to the fact that Stephanie could wash dishes but she takes a very long time to do so. However, she cannot do house chores like cleaning the floor and applying polish.

*“She makes such a mess – to a point that you just tell her to stop.”*

#### **8.4.1.17 Social support**

Anna reported that she has inadequate support. She would like to have someone who could take Stephanie for a few days, so that she can take a break. Anna mentioned that she gets limited support from her older sister but her mother does not want Stephanie to visit her. Stephanie’s father and the previous in-laws do not help her.

*“My mother does not want to stay with Stephanie. She says that she is troublesome.”*

#### **8.4.1.18 Marital support**

When I first met Anna in 2011, she and her husband had been married for 18 years. Her husband was not emotionally supportive and they were experiencing marital problems. Their relationship was dysfunctional, as he refused to help take care of Stephanie. In 2013 the situation had deteriorated even further, as she reported that she was no longer able to cope with their marital problems. Her parents-in-law were also not supportive. She spoke about the problems in her marriage and the responsibility placed on her for taking care of Stephanie in an overwhelmed and hopeless manner. She mentioned that she had constant headaches and physical pains.

This concurs with Caldwell and Taylor’s research (1988). They gathered information from 12 families and found that parents had different views on how the birth of the child impacted on their marriage. Some parents suggested that it caused marital problems. Whitman (2006) also found that parents of children with PWS reported higher levels of parental and family problems. They further highlighted that having a child with PWS places further stress on marriages.

#### **8.4.1.19 Government support**

Stephanie is a recipient of a disability grant, and the two other children also get social grants from the government.

#### **8.4.1.20 Future plans**

Anna was pessimistic. She did not seem to have any future plans for Stephanie.

*“What plan? ... What would be a plan, I foresee problems.”*

#### **8.4.1.21 Sexual challenges**

In 2012, Anna reported that a teacher had informed her that Stephanie was talking about sexual activities with her friends. When confronted about this, Stephanie informed them that she was raped. Anna could recall that she saw that her underwear was bloody while she was doing her washing. They took her to the doctor, who confirmed that she had been raped. A case was opened and they were referred to another psychologist for counselling but the psychologist did not contact them. In 2013 Anna reported that Stephanie was allegedly raped again. In 2014 Anna mentioned that there were no significant behavioural and emotional changes observed, following both alleged rape incidents. Recently, when Stephanie sees a man she flirts with him, claims that he is her boyfriend, and tells people that she has a boyfriend.

#### **8.4.1.22 Skin picking**

In 2012, Anna reported that Stephanie had pierced herself with an earring. She also mentioned that Stephanie was once admitted to hospital because she had developed a septic wound which resulted from skin picking. When Stephanie presented in 2017, she had observable serious wounds around her chin as a result of skin picking (see figure 8.1, below). Anna mentioned that when Stephanie has a sore – she picks at it until it becomes bigger. Even when the sore is healing she will start the process all over again, and when she is told to stop she continues.

*“When you tell her to stop, it is as though you telling her to continue.”*

*“It is better she does not have diabetes – otherwise it would have been worse for her.”*



According to Gourash and Foster (2005) the skin picking behaviour of PWS has a wide range of severity across different patients, and sometimes in the same patient over time. Some patients have occasional minor skin picking, while others maintain large open wounds as has been the case with Stephanie (see figure 8.1, below).



**Figure 8.1:** The extent of skin picking on Stephanie's chin and hands, and illustrating the brace she has to wear every day.

#### **8.4.1.23 Hobbies**

Anna reported that Stephanie likes playing with her doll.

#### **8.4.1.24 Psychological support**

In 2011, Anna needed assistance managing Stephanie's behaviour problems at home, and in 2017 she needed assistance managing her behaviour problems at school. The school needed assistance in terms of an intervention so that they could use this to manage Stephanie's behaviour.

*"Please help me and help the teachers at school."*

#### **8.4.1.25 Prader-Willi Syndrome Association of South Africa (PWSSA)**

Anna mentioned that the Association is more like a support group for them. She said that the PWSSA gets donations from individuals and organizations. When they attend the annual general meeting, they receive the annual financial statements. The

plan of the organization is to build a home for children and adults with PWS but they have been struggling to source funding. Anna mentioned that they do not get important information from the Association as they are often sent information through e-mails, but she does not have access to a computer and is computer illiterate.

#### **8.4.1.26 Knowledge about other PWS members**

Anna knew about the challenges Dikeledi was experiencing with Kamo as described in Chapter six. She also informed me about a family that took their adult daughter to a facility, but the daughter had to return home as they could not manage the behaviour problems. Anna knew about the death of Irene's husband described in chapter seven. This suggests that the PWSSA members share information with each other.

#### **8.4.1.27 Recommendations**

Anna recommended that the government should build a home for children and adults with PWS, so that parents can continue to work. If that fails, parents should get a grant that will assist them, as they cannot work while they have to constantly take care of the individual with PWS.

### **8.4.2 Themes identified from interviews with Stephanie**

During the interviews and sessions with Stephanie, she was cooperative. However, she struggled to understand some of the questions posed to her. She seemed to have a concrete way of understanding questions and statements. She also demonstrated perseveration related to certain topics. The following themes were identified from our sessions:

#### **8.4.2.1 Diagnosis**

Stephanie reported that her mother had told her that she had PWS from a young age.

*"My mother told me."*

*"She told me I have a disease called Prader-Willi Syndrome."*

#### **8.4.2.2 Impact of PWS on her life**

She mentioned that PWS has a negative impact on her life, as it affects her health.

*“You get sick ... I now have scoliosis, my back bends, and it gets out of shape.”*

#### **8.4.2.3 Food-related problems**

Stephanie reports that she gets upset when her mother does not give her breakfast. She also mentioned that she gets food from her mother.

*“I eat at home; I don’t starve.”*

#### **8.4.2.4 Behaviour problems**

Stephanie reported that she does not like being naughty. She mentioned that if she were to change anything in her life, she would stop bothering people. This suggests that she could be experiencing self-stigma. According to the stigma theory, self-stigma includes the apprehension of being exposed to the stigmatization, and also potential internalization of the negative beliefs and feelings associated with the stigmatized condition (Bos et al., 2013). She reported that she asks people for money and uses the money to buy snacks.

*“I ask people for money – it is my way. I am a hustler.”*

*“When I get home, I go [to] the shop and I buy a big Ultra Mel custard, chips, stock sweets and biscuits.”*

She reported that she only gets angry when other learners tease her, and when she is angry she keeps quiet. Stephanie further mentioned that the other learners tease her about her brace.

*“I get angry at people like Catherine.... When I am angry I just keep quiet.”*

#### **8.4.2.5 Treatment received**

Stephanie mentioned that she previously attended the endocrine clinic and currently is attending the spinal unit. She added that she is due for an operation, but she will not agree to a blood transfusion because her religion does not allow it. She also consulted with Dr. Duncan in the genetics clinic.

*"I am supposed to go for an operation – but if I will have a blood transfusion [and] I will not agree to the operation."*

*"I am a Jehovah's Witness. We do not do blood transfusions."*

*"I also consulted with Dr. Duncan."*

Stephanie was taking amitriptyline which she reports that she got from a dermatologist.

*"I am taking pink tablets that help my mind to calm down."*

#### **8.4.2.6 Milestone developments**

Stephanie mentioned that she had started menstruating and does not like it when she has a period. She often tells her teacher about her periods and asks for sanitary pads.

*"I am in trouble, am on my period ... it bothers me."*

#### **8.4.2.7 Siblings**

Stephanie stated that she gets along with her siblings. However, she experiences problems with Lucky because he beats her.

*"We are ok. But Lucky likes to beat me when I ask him to prepare food for me without any reason."*

#### **8.4.2.8 Social support**

Stephanie reports that she has a good relationship with her mother. She mentioned that her aunt is part of her support system, and that she visits her occasionally during the school holidays. Her father also visits them occasionally but she has not seen him in a while.

#### **8.4.2.9 Social life**

Stephanie reports that she has friends at school. They play swing and driving cars. She has one friend in the neighbourhood, and they like playing house. Stephanie claims to have a boyfriend.

*"His name is Aubrey – he is my man."*

*"I like playing school and house."*

Goff (2006) mentioned that adolescents with PWS desire the same things that other teenagers want: friends and close relationships. They can, however, carry this desire to extreme lengths in their search for a boyfriend or girlfriend including obsessing over a particular individual.

#### **8.4.2.10 Adaptive functioning**

Stephanie is able to bath and dress herself.

*“I dress myself.”*

#### **8.4.2.11 Sexual challenges**

Stephanie reported that she was allegedly raped twice. She mentioned that she was not bothered by the incidents and that she is coping well. She spoke about the incidents in a superficial manner which might suggest that they did not have a significant impact on her.

#### **8.4.2.12 Medical problems**

Stephanie reported that she sometimes experiences a burning sensation in her stomach. She also stated that she has scoliosis.

#### **8.4.2.13 Skin picking**

Stephanie mentioned that she bites her nails in order to keep them clean (see Figure 8.1, above).

*“I remove my nails, so that they do not get dirty and have germs.”*

#### **8.4.2.14 Government support**

Stephanie currently receives a disability grant.

*“I get a disability [sic].... They use it to buy me food.”*

#### **8.4.2.15 Schooling**

In 2011, Stephanie mentioned that she was expelled from her previous school. She was in a new school then and she did not like it, she wanted to study in Gezina. She currently (2017) reported that she was experiencing problems at school because she

has to move to another class but she does not want to move because she gets bored in the other class and the class teacher beats them. The current class is better, because she gets an opportunity to learn how to read. Stephanie insists on staying in the same class. She tried to convince me that in the current class they learn whereas in the other class they play. She continuously emphasized this point. Stephanie also mentioned that in the current class they have different educational toys.

*"I am not going to Ms Jennifer's class – I am going to my teacher's class."*

*"It is boring in there, they beat us."*

*"I want to learn how to read."*

*"It is boring in that class – I find this one better."*

*"It is boring, they play with toys and watch television."*

*"We don't go there to play, we learn."*

*"I want them to add me on my teacher's register. That is all I want, that is all I want."*

Stephanie added that her teacher gives them food and when it is her birthday she brings them cakes and snacks. This could possibly contribute to her resistance to moving from the class.

*"She brings cake, chips, peanuts, juice, and shares with us."*

Contrary to what Anna had said about chores, Stephanie reports that when she is at school she cleans and polishes the floor in her class.

*"I clean, I polish, I move desks when I am cleaning – even my teacher can tell you."*

*"I polish the floor until it shines."*

Stephanie mentioned repeatedly that she listens to her teacher and that she follows her instructions when Anna is not there.

*"I listen to my teacher, when she says: 'do this', I do it."*

*"I behave myself when you [are] not there, seriously ... you must ask my teacher."*

*"You must ask my teacher – when you are not there I listen to her."*

*“You must ask her in front of me. I am telling you, she will tell you that I listen to her.”*

This was evidence of perseveration and Stephanie did this during different sessions, whenever she wanted to emphasize her point.

During a different session, after interventions were implemented and Stephanie had moved to Ms Jennifer’s class, she emphasized that she would eventually like to go back to Mrs Maphosa’s class. Once again, perseveration was noted:

*“What I want is for you to tell the principal to take me back to Mrs Maphosa’s class.”*

*“Mom! Mom! What I want is for you to tell the principal to take me back to Mrs Maphosa’s class – do you hear me?”*

*“That’s all I want; are you going to tell her?”*

#### **8.4.2.16 Future plans**

In 2012, Stephanie reported that she would like to become a paramedic and to drive an ambulance when she finishes school. She wants to get married to a Chinese man and to have 10 children. In 2017, she still maintained that she would like to work as a paramedic and to have 10 children, but she now wanted to get married to a white man. She seems to fantasize about having her own family.

*“I would like to be a paramedic, they get paid a lot of money.”*

She mentioned that when her parents die, she is going to stay with her teacher.

*“I will stay with my teacher. She stays in The Orchards.”*

#### **8.4.2.17 Recommendations**

Stephanie recommended that parents should buy their children food.

*“I will tell them bring your children food – do not starve them.”*

### **8.5 Collateral information from class teachers**

In 2011, a school feedback report (to be completed) was given to Ms. Thabethe, the class teacher at Neo Mathabe special school. The aim was to gather information on Stephanie's behaviour and performance in the school environment. The class teacher reported the following:

- Stephanie's school performance was gradually improving.
- She had respect for educators and other elderly people at school including the cook, assistant teacher, and transport driver.
- She participated fully in school activities with other learners in class and outdoors.
- Stephanie got angry with her peer group, because they teased her a lot.
- She stole a lot even at home. As a result, the mother worked together with the teachers to find a solution.
- Stephanie became emotional and screamed or even cried.

In 2017, Mrs. Maphosa, the current teacher at Neo Mathabe special school, came to the hospital to give me information on the behavioural problems they experience with Stephanie. They needed suggestions on how to manage the behaviour. She mentioned the following problems:

- Stephanie does not want to stay in her registered class, she wants to be in her class. She does not like her registered teacher and throws tantrums when she is forced to be in her class.
- When she doesn't want to do something like writing, she simply refuses to do it.
- She cannot sit still in class.
- Stephanie cannot concentrate on a given task for more than 5 minutes.
- She sometimes throws terrible tantrums. During the tantrum she shouts, screams, swears, throws herself on the floor, and sometimes runs away. When she has a tantrum, she brings the whole school to a standstill.
- Stephanie sometimes locks herself up in the bathroom.
- When she goes to the toilet she spends a long time there.



- Mrs. Maphosa has to accompany Stephanie when she goes to the toilet, in order to prevent her from locking herself in.
- She steals anything she lays her hands on, such as her food, other learners' food, books and stationery, and she hides the stolen items in her brace or in her underwear.
- Stephanie masturbates in class in front of other learners.
- She undresses in class and show other learners her private parts.
- She talks to other learners about sex.
- Stephanie has an imaginary boyfriend.
- She does not respect others, and she swears at other learners. Some learners fight with her when she swears at them.
- She skin-picks and this becomes so intense that she even uses scissors – which results in more injury to the skin.
- Stephanie steals from the neighbours of the school.
- Mrs. Maphosa described Stephanie as lazy, but when Stephanie wants to impress her, she will take a broom and sweep the floor but this does not happen often.

Mrs Maphosa has resorted to the extreme measure of using handcuffs on Stephanie, to prevent her from skin picking and masturbating in class. Other teachers feel they cannot be controlled by a child.

## 8.6 Interventions

### • ***Behaviour therapy***

According to Corey (2009) behaviour therapy practitioners focus on observable behaviour, current determinants of behaviour, learning experiences that promote behaviour change, tailoring treatment strategies, assessment, and also evaluation. The aim is to define the problem, identify what needs to be changed, assess the effectiveness or ineffectiveness of the current intervention, and devise intervention strategies. This is supported by Grant and Evans (1994) who advised that when facing a behavioural problem, one of the first things is to identify what is to be changed. They further emphasized that it is important for the behaviours to be clearly defined. These were implemented with this particular family.

## **2011 to 2014 sessions**

- ***Identifying the problem and what needed to be changed***

When we first consulted with the family in October 2011, Anna reported the following problems:

- Stephanie steals food.
- She asks for food and money from people she meets.
- She eats a lot.
- She locks herself in the toilet.
- Stephanie baths for a long time.
- She defecates at various places in the school premises.
- She swears a lot.
- Stephanie does not listen when she is disciplined.
- She steals from the shops.

- ***Anticipatory planning***

We devised strategies to help Anna to be pro-active and to prevent the behaviour from happening. This is supported by Whitman (1995) who said that behaviour management must focus on preventing the opportunity for expression of such behaviours, rather than depending on the development of internal cognitive controls.

The strategies devised were:

- Instruct Stephanie not to ask for anything when they arrive at a place or tell the people around them not to offer Stephanie food or entertain her requests for food or money when she asks for such.
- Do grocery shopping or go to the shopping mall when Stephanie is at school in order to prevent her from stealing from the shops or asking people there for money and for them to buy her things.
- Lock all food items in the shack to ensure that Stephanie does not have access to food, and in the process, prevent her from overeating. This is in line with IPWSO (2013) that control of food-related behaviours is complex, but centres on strategies to limit access to food. These include consistent limit setting, locking cabinets and the refrigerator at home, limiting exposure that could make the child think about food, and the avoidance of environments with available food.

- Be firm and consistent with all the above.

By March 2012, Anna was proactive in her management of Stephanie's behaviour: She reported improvement in Stephanie's behaviour and the improvement was maintained throughout the year. However, Anna complained that managing the behaviour by being proactive was stressful for her, as she had to monitor Stephanie's every move.

In January 2013, due to the stressors associated with her marital problems, Anna was unable to concentrate on and pay attention to pre-empting Stephanie's behaviours like before. By June 2013, improvement in Stephanie's behaviour was still reported. However, there were problems when Anna was pre-occupied with and distressed by her marital problems and she could not be proactive in managing the behaviour. Stephanie would then take advantage of the situation. I tried to support Anna with individual psychotherapy. This is in line with Cassidy and Driscoll's (2009) research findings that highlighted that it is important to not only focus on the diagnosed individual, but also to provide the necessary intervention to the primary caregivers as they are confronted by multiple stressors. Whittington and Holland (2010) also stated that families of people with PWS are subject to stress and that 70% of the mothers have high levels of stress which require psychological counselling.

Following her own psychotherapy in October 2013, Anna became emotionally stable, and, as a result, she was able to pre-empt the behaviours which then resulted in greater improvement in Stephanie's behaviour. The swearing had improved, Stephanie no longer had the opportunity to ask people for food and money, and she was no longer being taken to the shopping mall. The improvement in Stephanie's behaviour was maintained. In 2015 Anna stated that the interventions around ensuring food security, pre-empting the behaviour, and providing consistency in the environment, were working effectively.

In 2017, Anna gave feedback on the interventions which had previously been implemented. She mentioned that since she attended psychotherapy, her stress

levels had decreased. Anna was now able to focus on Stephanie's behaviour even though it tends to be emotionally exhausting and time-consuming for her. She still locks food in the shack, but sometimes the siblings forget to lock the shack and only then does Stephanie get the opportunity to steal food. Anna still leaves her at home when she goes shopping. When she takes Stephanie in public Anna warns people in advance before Stephanie asks them for food. Most of the behaviour challenges they had at home are still managed effectively, and they were now dealing with different behaviour problems.

### **2017 sessions**

- ***Identifying the problem and what needs to be changed***

Anna reported the following problems at home:

- Stephanie drinks medication she has access to even if it belongs to other people.
- She hides her siblings' books and stationery or gives the items away.
- Stephanie still does not listen when she is disciplined.
- She interrupts Anna when she is having conversations with other people.
- She swears at Anna and the siblings.

Anna and Mrs. Maphosa reported the following problems at school:

- Stephanie refuses to go to her registered class.
- She throws temper tantrums.
- She locks herself in the toilet.
- She runs away from school.
- Stephanie steals from other learners, teachers, and also neighbours.
- She swears at both teachers and other learners.
- She masturbates in class in front of other learners.
- She talks to other learners about sex.
- She picks her skin using sharp objects

.

- ***Punishment***

Anna beats Stephanie or deprives her of snacks or does not give her breakfast, which seems to be ineffective as it had not yielded positive outcomes. When this happens, Stephanie throws a tantrum. This confirms the statement that parents

should be advised that offering food as a reward or withholding food as a punishment, is nearly always counterproductive, and should be avoided (McCandless et al., 2011).

- ***Anticipatory planning***

The following were suggested:

- When the siblings finish their homework, they should lock their school bags in the shack to prevent Stephanie from stealing their books and stationery.
- To avoid a situation where Stephanie is taking and drinking medication that she finds in the house, the long-term plan is to get a locking cabinet in the bathroom, where Anna will store the medication. Temporarily she will remove all the medication from the house and lock it in the shack.

This concurs with the assertion that good management means always anticipating tomorrow, next week, new situations, changed plans, and the like (Whitman, 1995).

- ***Pivot tool***

When Stephanie interrupts Anna or swears at them, Anna will ignore her and the siblings will do the same. This approach is supported by Whitman and Jackson (2006) who described the praise and ignore (pivot tool) which illustrates that when inappropriate behaviour occurs, the strategy is to pivot attention away from the problem behaviour to another task.

### ***Managing behaviour problems at school***

- ***Positive reinforcement***

Positive reinforcement involves the addition of something of value to the individual (e.g. praise, attention, money, food) as a consequence of certain behaviour. When the goal of a programme is to decrease or eliminate undesirable behaviours, positive reinforcement is often used to increase the frequency of more desirable behaviours, which then replace undesirable behaviours (Corey, 2009).

Stephanie found toy cell-phones in my office which were still sealed in their wrapping. She seemed to be interested in one, and she opened it without permission. I took advantage of the interest and informed her that I would give her the phone when she comes for a follow-up session provided that she moves to her

registered teacher's class. I also used Mrs. Maphosa as a "reward": if she goes to the registered class she will be able to spend approximately 15 minutes with the teacher and get "special time" from her. Positive reinforcers are generally not difficult to identify, and reward systems that use small, short-term goals that progress to larger goals, are typically effective (McCandless et al., 2011). Within two weeks, Mrs Maphosa and Anna informed me that Stephanie had moved to the registered teacher's class. I gave her the toy cell-phone and she was very excited.

- ***Pivot tool***

In order to manage the swearing, I suggested that when Stephanie starts swearing at them they should remember that it is not personal, ignore her and focus on other activities until she stops. This is supported by Whitman and Jackson (2006) who described the praise and ignore or pivot tool, which illustrates that when inappropriate behaviour occurs, the strategy is to pivot attention away from the problem behaviour to another task.

- ***Psycho-education***

According to Lukens and McFarlane (2004) psycho-education is among the most effective of the evidence-based practices that have emerged in clinical trials and community settings. It is a professionally delivered treatment modality that integrates and synergizes psychotherapeutic and educational interventions. Psycho-education reflects a more holistic and competent-based approach stressing health, collaboration, coping, and also empowerment.

A psycho-education model therefore views the role of the psychological practitioner not in terms of abnormality, diagnosis, prescription, therapy and cure but rather in terms of goal-setting, skills teaching, satisfaction, or goal achievement (Authier, 1977).

Family psycho-education includes the teaching of coping strategies and problem-solving skills to families, friends, and/or caregivers to help them deal more effectively with the individual. The rationale for the psycho-education is that it reduces distress, confusion, and anxiety within the family, which may in turn help individuals to improve their lifestyle (Ahmed, 2004).

Psycho-education was done with the class teacher, and the following suggestions were made before Stephanie actually moved to her registered class:

- She should remain temporarily in Mrs. Maphosa's class to ensure minimal disruption for the rest of the school while trying to devise a solution.
- There should be rules in her class and they should be firm and consistent in ensuring that the rules are followed.
- Stephanie should be kept busy, in order to minimize disruptions, skin picking, and masturbation.
- Consistency, certainty and structure should be provided, which might lessen the anxiety, and possibly improve skin picking.

I also visited the school and did psycho-education with all the teachers. At the beginning of the session I asked them to rate their knowledge of PWS using the rating of 1 being no knowledge, and 10 being that their knowledge is good. The majority rated themselves zero which was below no knowledge, and only two rated themselves between one and two. The teachers really appreciated the information and felt empowered and educated. They even had empathy for Stephanie and their perceptions regarding the displayed behaviours changed. Chedd et al. (2006) stated that a school can benefit from regularly scheduled parent conferences and outside consultations with professionals familiar with PWS.

In a follow-up session, Anna gave the feedback that the teachers were now aware that controlling the environment was crucial. As a result, the principal made sure that the longstanding plan of building a fence around the school was implemented with immediate effect to ensure that Stephanie could not run away from school. Both teachers and parents need to provide extra security in the school and at home. This was true for the teachers at the school. This intervention also helped Anna, as they were no longer blaming her and placing responsibility on her for Stephanie's behaviour.

The skin picking also improved and the area (the chin) where she was skin picking was visibly improved (see Figure. 8.2 below compared to the right part of Figure 8.1). Based on this improvement, the skin picking could be said to be attributed to the

anxiety Stephanie was experiencing in the school environment. This could have been perpetuated by the instability, inconsistency, and changes imposed on her. Whittington and Holland (2010) stated that skin picking has been related to boredom and anxiety but objective evidence for this is difficult to establish. Therefore, what was observed with Stephanie might help confirm the possibility that skin picking is associated with anxiety.



**Figure 8.2: Stephanie's chin area, showing improvement in skin picking following the described interventions.**

- ***Referral to peadiatrics OPD and psychiatry OPD (Out-Patient Department)***

I referred Stephanie to Dr. Duncan who is in the peadiatrics OPD, in order for her to do the necessary blood work so that I could refer her to psychiatry, and also for her to prescribe some medication to manage the behaviour temporarily while waiting to consult with a psychiatrist. Dr. Duncan prescribed the atypical antipsychotic Risperdal (risperidone). Risperdal is a serotonin-dopamine antagonist, and is an effective antipsychotic medication (Sadock & Sadock, 2003). The teacher and primary caregiver gave feedback after two weeks of being on treatment. They reported that Stephanie was much calmer. She mostly sleeps in class but they feel this is better for them as she is no longer disruptive and is not displaying the previously stated behaviour problems. The behaviour in class is now controlled.



According to Whitman and Jackson (2006) the use of psychotropic and neuroleptic medications (designed to alter behaviour, mood, or thought by targeted action on brain chemistry) is increasingly advocated for supplementing behaviour-management strategies or as a primary intervention in the case of severe emotional or cognitive disruption (e.g. depression, psychosis) which was the case with Stephanie. Something was needed to supplement the behaviour-management techniques, as their effectiveness seemed to be short lived. Many parents have mentioned that the need to medicate is for the convenience of the service provider (e.g. group home staff, teacher) rather than the needs of the person with PWS (Whitman & Jackson, 2006). This was also the case with Stephanie, as the use of Risperdal was to ensure that the behaviour problems, which were observed to be out of control were managed effectively, and that the teachers and primary caregiver manage her better and cope better in terms of taking care of her.

After six months, a brief follow-up was done with the family. Stephanie appeared to have lost weight and Anna attributed this weight loss to a strict food regime and ensuring that the shack is locked at all times. Anna reported that since they implemented the behaviour techniques as well as ensuring that Stephanie adheres to the treatment she receives from the psychiatrist her behaviour has been better controlled, she is calmer and there is a lesser need to constantly monitor her. Due to this change, Anna is now job seeking and she was recently invited to a job interview.

The teachers at her school understand her behaviour better and they have modified the school environment by building a wall fence around the school and ensuring that food access is limited. The teachers also confirmed that Stephanie is calmer at school – she no longer locks herself in the toilet or try to run away, does not masturbate in class anymore and there have not been reported incidents of her stealing or swearing. Subsequently the teachers are coping better with managing her at school.

## **8.7 Conclusion**

This chapter demonstrates that it is not easy to clinically diagnose PWS: It can be misdiagnosed, and as a result Stephanie had the wrong diagnosis for seven years. This case also illustrated that Stephanie was experiencing typical characteristics of individuals diagnosed with PWS. The behaviour problems displayed by Stephanie affected everyone around her. The teachers and her mother struggled for a long time to manage these behaviour problems.

This case has also revealed that taking care of an individual with PWS is time-consuming, and is physically and emotionally exhausting for the primary caregiver as she must monitor her daughter around the clock. This does not only have an impact on the primary caregiver but also on the siblings and teachers. Based on their lived experiences, Anna seems to be deprived of an adequate support system. Anna was in fact in dire need of social and financial support.

The government grants which the children receive have played a crucial role in this family. It is the only source of income which sustains this family. One cannot underestimate the impact that unemployment has had on Anna, because she cannot fully provide for her family as a result. The unemployment could be attributed to the fact that Anna has to constantly take care of Stephanie, and she is therefore unable to get a full-time job.

Despite her PWS diagnosis, Stephanie seems to have needs (just like any other adolescent) such as sexual desires and fantasies of having a boyfriend and a family. However, Stephanie presents these needs inappropriately, and, as a result, they had to be managed.

This case also reveals that after there had been significant changes in Stephanie's life, the behaviour problems and skin picking were exacerbated. This could possibly be associated with inconsistency, instability and introducing changes in Stephanie's life which made her feel anxious. This is so, because when the school environment was more accommodating and ensuring there was consistency and stability and the skin picking diminished.

Interventions like positive reinforcement, the pivot tool, anticipatory planning, psycho-education and medication, proved to be effective in managing some of Stephanie's behavioural problems both at home and at school. The teachers and the primary caregiver were empowered during this process, and were involved in developing the treatment plan. This case also demonstrates once the behaviour problems are effectively managed, the teachers and primary caregivers coped better and the primary caregiver was able to start the job seeking process.

## **CHAPTER 9**

### **CONCLUSIONS, EVALUATIONS AND RECOMMENDATIONS**

#### **9.1 Introduction**

The present study was inspired by my involvement with Stephanie, a child with PWS who had been manifesting behaviour problems. At the time, I had no knowledge of PWS, neither did my colleagues and other healthcare professionals. This lack of knowledge among healthcare professionals, inadequate literature on the psychological impact of PWS, and limited studies conducted in South Africa, sparked my interest. This was my first step in terms of deciding to conduct the present study.

In addition to genetic and physiological challenges and also cognitive impairment, individuals with PWS display a range of behavioural problems that become noticeable in early childhood and increase during adolescence and adulthood.

#### **9.2 Research objectives and the implementation thereof**

The primary aim of this study was to learn about the experiences and challenges of the individual diagnosed with PWS. The behavioural problems tend to impact those around them, especially the primary caregiver which often results in the caregiver being unable to cope and needing counselling. Thus, the secondary aim of this study was to build up knowledge on the primary caregiver's experiences and challenges. The above aims were achieved by conducting semi-structured interviews with both the diagnosed individual and the primary caregivers as well as by using observations.

Existing research on effective behavioural techniques has been less extensive and hence the last aim of this study was to assess the effectiveness of existing interventions. This was achieved in two ways. First, the primary caregivers had previously implemented certain interventions to try and manage the behaviour problems, and the effectiveness or lack thereof was based on their experiences. Secondly, I conducted additional session/s following the semi-structured interviews, and in these sessions tailored intervention/s based on their challenges and experiences were suggested. These were aimed at easing their challenges and

improving their day-to-day functioning. The families were given approximately two weeks to a month to implement the intervention/s and feedback was given on the effectiveness or ineffectiveness of the intervention.

This study was undertaken by conducting qualitative research. In order to generate a comprehensive understanding of the participants, collective instrumental case studies were used, making use of participatory action research, ethnography and auto-ethnography elements. The data were gathered during semi-structured interviews and were organized around certain topics and common themes that emerged in each case study.

In this concluding chapter, I present a clinical description of PWS, and discuss the impact of PWS on the diagnosed individual, the primary caregivers, and on the school environment. I also share the psychological insights I have gained through my research. Then, I present some of my personal reflections, recommendations, and evaluations of the strengths and limitations of the study. Lastly, I make some suggestions for further research.

### **9.3 A clinical description of PWS**

The findings of this study showed that PWS similarly affects individuals from different races, genders, and socioeconomic status. These findings concur with previous research (Hurren & Flack, 2016; McCandless et al., 2011; Sinnema et al., 2011).

The participants in this study had observable dysmorphic features, small feet and a short stature. Only one participant appeared taller in this study, and she was of normal height compared to her peers. Three of the participants were observed to be obese whereas two appeared to be of normal weight.

The above findings support observations by Ho and Dimitropoulos (2010), Sinnema et al. (2011) and Hurren and Flack (2016) that individuals with PWS are typically short, obese, possess small hands and feet, and have dysmorphic features like a narrow bifrontal diameter, full cheeks, almond-shaped eyes, and a small mouth with downturned corners and a thin upper lip.

In addition to the above clinical observations, an interesting finding was that research participants had an observable gait abnormality, which is not widely documented in the literature. However, research by Cimolin et al. (2010) and Cimolin et al. (2017) found that in PWS, gait disorders are common, and are characterised by different gait strategies which tend to progressively worsen as the clinical picture advances, severely limiting the patient's quality of life. The results showed that the individuals walked more slowly, had shorter stride length, lower cadence, and a longer stance.

The majority of the participants did not cry immediately after delivery. These findings could not be verified with the literature reviewed. The literature only states that in infancy hypotonia is characterised by weak crying (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010) which was the case with only one participant, whose crying was reported to have been weak and soft after delivery.

The research participants had poor muscle tone (hypotonia) and gained muscle strength at varying rates ranging from two weeks to two months. This is similar to the findings in the literature, that hypotonia in infancy manifests with poor reflexes (Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010).

When the research participants were babies, they also slept excessively and would not wake up even to feed. The primary caregivers had to wake them up every two hours in order to feed them. This finding concurs with the research findings of Cassidy and Driscoll (2009) and Honey (2010) who stated that sleep problems are commonly observed and are reported to be one of the characteristics in individuals with PWS. Infants with PWS do not spontaneously demand feedings, and rarely wake up to feed.

Hypogonadism, which is another indicator of PWS, was reported in only the male participants, where it presented as under-developed testes and genitals at birth. This concurs with previous findings that under-developed sex organs (reflecting hypogonadism) are another indicator of PWS; this has been observed in both sexes (Angulo et al., 2015; Cassidy & Driscoll, 2009; Cassidy et al., 2012; Hurren & Flack, 2016; McCandless et al., 2011). What differed in this study, was that hypogonadism was not evident in the female participants.

All the participants had feeding problems as babies; they could not suckle and different techniques were used to feed them. Some were fed through feeding tubes, one was fed through a feeding cup, and another was fed through a spoon for a period ranging from two weeks to two months. This is in line with the literature that reports that assisted feeding through a dropper or spoon, a feeding tube and/or special nipples, and with increased feeding times, are necessary for a period of time, usually weeks to months to assure adequate calorie intake (Cassidy, 1988; Cassidy & Driscoll, 2009; Cassidy et al., 2012; Ho & Dimitropoulos, 2010). When the phase of feeding problems passed, the research participants started gaining weight.

This study revealed that the diagnosis of PWS was offered at different ages. For instance, some participants were diagnosed within a few weeks, and others within two months. Whittington and Holland (2010) reported there is now a generation of children who have been diagnosed with PWS within days or weeks after birth. Bar et al. (2017) found that the mean age at diagnosis was 18 days.

However, the diagnosis of others was delayed as it was only given after three years and seven years for other individuals. These results concur with Christianson et al.'s (1998) findings in South Africa, that the clinical diagnosis is considered to be difficult because many features are subtle or non-specific, and others change with age. Similarly, Caldwell and Taylor (1988) found that at the time of birth a diagnosis of PWS was extremely rare; infants needed to demonstrate the phenotypic characteristics: some were misdiagnosed or were not diagnosed at all.

Cassidy and Driscoll (2009), Driscoll et al. (2016) and Hurren and Flack (2016), all stated that children with PWS experience delayed milestones including gross motor and language delays. This was evident with all the research participants, in that sitting, crawling, walking, and speech were delayed. The speech difficulties with most of them seemed to persist except for one participant whose speech is now clear and audible.

The speech difficulties in individuals with PWS include poor speech-sound development, reduced oral motor skills, and language deficits. Language problems also include deficits in vocabulary, grammar, morphology, narrative abilities, and

pragmatics (Lewis, 2006). Similarly, the speech of the participants in this study was unclear and inaudible at times. They struggled with pronunciation of some words and their language expression was poor, and they had difficulty with elaborating on their answers except for one participant. In addition, one participant spoke in a hoarse and deep voice and had hyper-nasal speech. This presentation concurs with the view of Lewis (2006) who found that voice characteristics reported for individuals with PWS include a high-pitched voice, harsh/hoarse voice quality, inadequate vocal intensity, and hyper nasality.

Based on the above clinical features the participants received treatment from different healthcare professionals such as those in speech therapy, physiotherapy, occupational therapy, and also a dietician. The findings in this study further demonstrate the importance of a multidisciplinary team in the management of individuals with PWS. This is similar to the suggestion made by Cassidy and Driscoll (2009) that children with PWS should receive early intervention (including physical, occupational, and speech therapies).

Some participants also received psychiatric and neurology treatment, in order to manage medical conditions like febrile convulsions, depression and behaviour problems. This finding supports the research findings of Whitman and Jackson (2006) who found that the use of psychotropic and neuroleptic medication is increasingly advocated to supplement behaviour-management strategies or as a primary intervention in the case of severe emotional or cognitive disruption.

In the literature, many parents mentioned that the need to medicate is for the convenience of the service provider (e.g. group home staff, teacher) rather than the needs of the person with PWS (Whitman & Jackson, 2006). This was also the case with four of the research participants, as the prescription of the medication was to ensure that the behaviour problems which were out of control, would be managed effectively and the teachers and primary caregiver coped better with taking care of the participants as a result.

Three of the participants were also on GHT, one for a short period and the others for longer periods. The other participants did not receive it at all. All the individuals who



received GHT got it for free from government hospitals which was significant because for one who was born in a private hospital it had to be funded privately, and being very costly forced them to stop. This compromised growth as a result. These findings are in line with those of Goranson (2011) who found that while some children with PWS got treatment, others were denied treatment because it could not be proven they had a growth hormone deficiency. Even if the doctor prescribed it, the family health insurance plan would not cover the cost, because it was considered experimental treatment for children with PWS. Therefore, because of the cost of medication the promise of a better life for their child was out of reach. This finding suggests that it is crucial for medical aids in South Africa to consider classifying GHT as a chronic medication for individuals with PWS.

McCandless et al. (2011) and Whittington and Holland (2010) stated that with growth hormone supplementation, individuals with PWS may have normal growth trajectories and a final height comparable with parental height. However, this proved to be the case with only one of the research participants in this study. The reason for this is unclear, and further research is warranted.

It was common with male research participants that their milk teeth did not come out naturally, and they had to be extracted. I could find no report of this in the literature, and thus this is an interesting finding and contribution to existing knowledge.

Based on the findings of this study, healthcare professionals seem to be concerned about diabetes as participants were screened for diabetes at every check-up and one was even prescribed glucophage. Butler et al. (2006) reported that PWS is associated with a tendency to show an increased risk for the development of diabetes mellitus and that type II diabetes is in 25-30% of PWS adults who become morbidly obese. The results of this study demonstrate that despite the age of the individual with PWS screening for diabetes in the South African context is prioritized. Thus far the majority of participants in this study did not have diabetes.

Another feature of PWS which can be reported from the research participants, was skin picking and a probable high pain threshold. With most participants, this was minimal, but in one it was severe. The skin picking could be attributed to the high

pain threshold. It was reportedly not painful even when this was done until blood came out from the wound. This concurs with the report by Gourash and Foster (2005) who found that the skin picking behaviour of PWS has a wide range of severity from patient to patient, and sometimes in the same patient over time. Some patients have occasional minor skin picking while others maintain large open wounds.

According to Whittington and Holland (2010) skin picking has been related to boredom and anxiety. However, objective evidence for this is difficult to establish. The findings of this study possibly substantiate this suggestion. It was evident that when Stephanie was overwhelmed by the changes occurring in her school environment, the skin picking became severe and could thus be attributed to anxiety. During the interviews held with the primary caregivers and individuals with PWS, whenever the focus of the questions was on the primary caregiver, the PWS individuals started skin picking which means that it is probably associated with boredom.

#### **9.4 The impact of PWS on the diagnosed individual**

Food-related problems were a challenge for all the participants, but the onset differed and ranged from early childhood, to late childhood and late adolescence. The research participants were all stealing food and were accepting food from other learners at school. One participant would steal uncooked, frozen foods. Two participants stole money, sold items and asked people for money all in order to buy food; otherwise they simply asked people for food. The findings of this study confirm Gourash et al.'s (2006) finding that food-seeking behaviour may include foraging for food in and out of the home, and also consuming spoiled, raw, frozen, or otherwise inedible foods. However, in the current study, the consumption of spoilt or inedible foods was uncommon.

An unexpected finding was that with Tshepiso, the food-stealing behaviour was resolved, and she is now disciplined and has self-control around food even when she is unsupervised.

Three research participants had satiety problems. A surprising finding in this study was that despite having food-related problems, two participants did not have satiety problems (hyperphagia). They could tell when they were full and would stop eating. This was evident in that they had normal weight compared to other participants. This suggests that although all the individuals are diagnosed with PWS they presented differently. It was speculated that all the research participants would have hyperphagia, as it has been documented as a central feature of the syndrome (Butler et al., 2006; Cassidy & Driscoll, 2009; Honey, 2010; IPWSO, 2013; King, 2008; Whittington & Holland, 2010) and a given that they stole food. However, this research demonstrated that not all individuals with PWS have satiety problems.

King (2008) suggests that hyperphagia begins in late childhood, Honey (2010) wrote that it becomes evident as early as the age of two years, while Butler et al. (2006) reported that it begins by the age of eighteen months. It was thus of interest to note that Reba's hyperphagia began at seven months. This finding is significant and adds valuable information to the literature.

Multiple behaviour problems were reported throughout the literature reviewed (Butler et al., 2006; Cassidy & Driscoll 2009; Ho & Dimitropoulos, 2010; McCandless et al., 2011). The participants in this study also presented with different behaviour problems. For instance, they became frustrated and anxious when there was a change in routine and a lack of structure. Most had temper tantrums/anger outbursts. When they had temper tantrums they would say negative things, use angry words, and would tend to apologise afterwards. The participants were also stubborn and struggled to take orders from certain individuals in their lives. Other participants demonstrated manipulative tendencies, were argumentative, and had aggressive tendencies.

Participants also presented with obsessive-compulsive behaviours, which ranged from building puzzles, focusing on one task at a time and completing it before moving on to the next one, through to always buying more than one item at a time, organizing objects into a certain pattern, copying paragraphs from magazines, and obsessing over certain events and objects. Perseveration was observed and reported, as the participants tended to focus on and talk about certain topics for long

periods, ask many questions, and when they are passionate about something they would emphasize their point in relation to it.

Another notable finding was that three of the participants were reported to have encopresis and one also had enuresis. The encopresis in one participant improved but it remains persistent with the others. This is rarely referred to in the literature; however, Equit, Piro-Hussong, Niemczyk, Curfs, & von Gontard (2013) investigated the rate of elimination disorders and behavioural symptoms in persons with Prader-Willi and Fragile-X syndromes, because very few studies had addressed elimination disorders in persons with specific syndromes. They found that 29.3% of persons with PWS had at least one elimination disorder. Three of the research participants had severe behaviour problems which needed psychological and psychiatric interventions. Of note was that these three participants were also the ones who had elimination disorders. For two of them, these behaviours had a significant impact on their schooling, and it therefore required intervention at school and at home.

Despite their behavioural problems, it is important to note that people with PWS have many positive characteristics. They are known to be friendly, sociable, kind and caring and many have a wonderful sense of humour (IPWSO, 2013; PWSA UK, n.d.). In line with these assertions, participants in this study presented as friendly and warm, and some of them were also spontaneous and talkative.

This study also found that participants had different hobbies but all their hobbies were solitary activities. This concurs with the observation that people with PWS tend to withdraw into solitary activities rather than activities with their peers (Whittington & Holland, 2010). In addition, all participants preferred to play with younger children compared to their peers, and one of them also preferred older children. This behaviour corresponds with what is mentioned in the literature that because the social cognition of persons with PWS may also be impaired, most people with PWS have difficulties relating to their peer groups and often prefer to be with older or younger groups (Whittington & Holland, 2010). Of note, and which is not mentioned in the literature, is the fascination of female participants with marriage and children.

Participants in this study demonstrated some adaptive skills, in that they could function independently and were able to take care of their personal hygiene. Others had secured employment, but one could not maintain employment due to emotional outbursts and the inability to take orders. This concurs with the findings of Drago (2006) and Kazemi and Hodapp (2006) who found that behavioural characteristics of the syndrome frequently lead to job terminations.

Based on the findings of this study, there seems to be a link between the physiological challenges the participants experienced from infancy and behaviour problems particularly those related to the inability to maintain emotional control. This could be further explained by child-developmental theories, as described by Calkins, Perry and Dollar (2016) who reported that the first two years of an infant's life are characterized by growth in physical, psychological, and social skills. One significant area of growth in early development is in infants' self-regulation. Self-regulation has been defined as an infant's ability to control emotions and behaviours in order to cope effectively with environmental demands across a variety of contexts.

The ability to regulate at these levels emerges, at least in a rudimentary form, during the prenatal period (e.g., sucking, head turning, reaching and touching, gaze aversion, self-soothing), and continues throughout early childhood. These activities and social interactions teach infants about contingencies between their arousal, behaviour and environmental responses, which is fundamental to their developing awareness of their own ability to control their behaviour and attention (Calkins et al., 2016) which then becomes impossible to achieve for individuals with PWS due to hypotonia.

### **9.5 The impact of PWS on the primary caregiver**

The primary caregivers of the research participants were mostly the mothers. They reported that during their pregnancy they had decreased foetal movements except for one participant. They also reported reduction and changes in the babies' movements. Some felt decreased foetal movement throughout their pregnancies, while others had normal foetal movements at the beginning of the pregnancy, but as the pregnancy progressed the movements were significantly decreased, almost to nothing. These findings concur with authors such as Cassidy and Driscoll (2009),

Cassidy et al. (2012), Ho and Dimitropoulos (2010) and Hurren and Flack (2016) who stated that in individuals with PWS hypotonia is prenatal in onset, and usually manifests as decreased foetal movement. This study highlighted differences in the foetal movements which is valuable information which has not previously been documented in the literature.

During the prenatal phase, of note was that the healthcare professionals could not explain decreased foetal movements but they still did not investigate. This finding demonstrates a lack of knowledge among the healthcare professionals. It was only during the postnatal phase that they started investigating and assisting the primary caregiver.

The primary caregivers had full-term pregnancies except for one with a premature birth. Three participants had caesarean sections for various reasons ranging from poor weight gain of the baby, through to breech position and the possibility of the umbilical cord entangling the baby. These results are similar to what was indicated by Cassidy and Driscoll (2009), Cassidy et al. (2012) and Ho and Dimitropoulos (2010) who stated that sometimes, due to hypotonia, there is an increased need for assisted delivery or caesarean section. However, two research participants had a normal vaginal delivery.

Caregivers received genetic counselling following the PWS diagnosis except for one participant whose child was initially misdiagnosed. During counselling they were given a poor prognosis and were informed about possible challenges like excessive weight gain, excessive appetite, behaviour problems, and intellectual disability. This concurs with the view of Caldwell and Taylor (1988) who mentioned that children with PWS were given a poor prognosis with a high probability of having significant cognitive limitations.

The participants reacted differently to the diagnosis: One family accepted it immediately, another family was in denial, and the other families struggled to accept it. This concurs with the research finding by Thomson et al. (2017) that carers of individuals with PWS reported high stress associated with the initial diagnosis. The primary caregivers' expectations for the future were lost when the primary caregivers

were told about intellectual disabilities. They realized that the dreams they had for their children were no longer achievable. They became pessimistic about the future. Living and raising a child with PWS was extremely difficult for all the families. Caregivers were all married, and as couples they gave each other support and relied on each other. This is consistent with the findings of Caldwell and Taylor (1988) who asserted that some parents indicated that the birth of a child with PWS strengthened their relationship as a result of mutual support. With time, however, the fathers got less involved in caring for the child, and three of the fathers died leaving the mothers as sole primary caregivers. One of the remaining couples divorced. This finding concurs with the research of Caldwell and Taylor (1988) and Whitman (2006), who found that some parents suggested that the birth of a child with PWS caused parental and family problems and placed further stress on the marriage.

This study also revealed that after giving birth to a child with PWS, the demands of caring for them and the fear of having another child with PWS led the primary caregivers to delay plans of having another child and one mother was even sterilized. It is only recently that some couples were considering having another child. The one mother who had children since the birth of the PWS child, had not planned to do so.

Because the individuals with PWS presented with food-related problems, as mentioned previously, the primary caregivers used different methods to manage this behaviour. They restricted food access by locking food away, cooked small portions to avoid having left-over food, monitored the individual, removed opportunities facilitating the behaviour, and set rules with consequences for when the PWS individual misbehaved. It is evident through these cases that the process of ensuring limited food access becomes the sole responsibility of the primary caregiver.

Due to the hyperphagia reported with some of the participants and food-related problems the PWS individuals tended to gain weight and it once again became the sole responsibility of the primary caregivers to manage/ameliorate the weight gain. They found different ways of managing the weight of the PWS individuals. They gave them small healthy meals, made them adhere to a strict diet, monitored the intake of sweets and snacks, and they also exercised together. These findings are in line with

suggestions made by IPWSO (2013) that it is imperative to stick to a strict schedule for meals and snacks, to limit portion sizes, and exercise is also strongly recommended.

What was significant in this study was that socioeconomic circumstances contributed negatively to attempts to try and manage the weight of individuals with PWS. The environmental modifications required in managing food-related problems were not possible due to inadequate space and financial constraints. They only bought food they could afford and not the food suggested by the dietician. This finding demonstrates that the socioeconomic status has an effect on type of food-management approaches used.

The lives of these families were all interrupted. This included disruptions of studies, job terminations, the inability to secure full-time employment, and working times were also affected. The caregivers dedicated most of their lives to taking care of their children with PWS. The primary caregivers found their sense of self being engulfed by the role of caring, and they lost touch with themselves: Not once did I hear caregivers talk about their own needs, because their main focus was taking care of their child with PWS, and also constantly monitoring them. This caring role proved to be strenuous, time-consuming, overwhelming and almost unbearable and led to stress and struggles with coping. Some primary caregivers needed psychological support to help them manage academic difficulties and behaviour problems at home and at school. These shared experiences concur with the findings of Whittington and Holland's (2010) study which found that 70% of mothers of children with PWS experience high levels of stress. Moss (2009) also found that heavy demands are made on parents and caregivers of individuals with PWS which usually lead to negative experiences and feelings. Thomson et al. (2017) also found that the carers' stressors included time constraints and physical and emotional tiredness.

This research study illustrated that primary caregivers experienced a need for external support, which would relieve them of caregiving duties for a period of time and allow them time to focus on something other than the role of being a caregiver. Some primary caregivers also deprived themselves of social support, because they did not disclose the PWS diagnosis of their children to their friends and other family



members. They subsequently did not have many people supporting them and/or offering empathy.

The findings of this study revealed issues around stigma. This was evident in how “others” blamed some primary caregivers for not disciplining their children, and how the primary caregivers took responsibility and blame for their children’s behaviour. The primary caregivers also tried to prevent self-stigma and felt stigma, by not disclosing the diagnosis to “others” or their children to prevent them from being pitied and treated differently. One caregiver only later disclosed her child’s encopresis and enuresis because she felt embarrassed.

All research participants were affiliated with the PWSSA. It became evident that the participants knew about each other’s challenges. This suggests that the PWSSA members are a close-knit group of people who knew each other and were, in an undocumented way, providing support to each other. They were able to give empathy to those members whose children had major behavioural problems. However, two of the families reported experiencing challenges instead of benefiting from the Association. This is because they were illiterate and could not understand English which was used during PWSSA meetings. Furthermore, they were unable to use computers, which resulted in them not receiving information circulated by email.

This study revealed that religion has the potential to play a significant role. In two of the families it affected how they managed PWS and how they coped with taking care of a child with the syndrome. It is significant that with one of the participants, religion influenced their decision not to undergo an operation, a decision which could potentially compromise the individual’s wellbeing and quality of life.

## **9.6 The impact of PWS on the school environment**

The participants seemed to have had different challenges with schooling but all the children were enrolled in special schools. Performance was poor and this inability was distressing for some primary caregivers, whereas others accepted that their children could not be educated in the usual way. This is confirmed in the literature which states that cognitive disability has been considered an integral part of PWS, and when PWS children start school they have multiple, severe learning disabilities

and also poor academic performance (Cassidy & Driscoll, 2009; Whitman, 1995; Whittington & Holland, 2010). The children were also teased and bullied by other learners at school.

In addition to learning difficulties, two participants presented with behaviour problems at school such as stealing from teachers and other learners, throwing tantrums, swearing at teachers and other learners, lying, being disruptive, refusing to be in class and taking instructions from teachers, difficulty in managing transitions, wandering around the school premises, undressing in front of other learners, encopresis, and also inappropriate sexual behaviour.

Subsequently some teachers were left feeling that the children were naughty and they were not disciplined. The teachers and principals also believed that these children's behaviours were disruptive to the day to day functioning of the school and it became practically impossible for them to focus all their attention on one child. Behaving in this manner also resulted in these children being expelled from some schools, while in other schools, teachers sought professional assistance from a psychologist.

## **9.7 Psychological insights**

- ***Anticipatory planning***

Anticipatory planning was proven to be effective when the primary caregiver plans in advance, prepares activities in advance to avoid sudden changes and delays, and negotiates with the individual if there are anticipated changes. This was also done by pre-empting the undesired behaviour before it happened, as well as by proactively reducing opportunities for an undesired behaviour to occur. The above is in line with the assertion that good management means always anticipating tomorrow, next week, new situations, changed plans, and the like. Whitman (1995) further stated that behaviour management should focus on preventing the opportunity to express such behaviours rather than depending on the development of internal cognitive controls (Whitman, 1995).

- ***Routine and structure***

Establishing a structured environment with a scheduled daily routine proved to be effective for the PWS individuals in the study, even though it required the primary caregiver to work hard at maintaining it. This is consistent with the views of Gourash and Foster (2005) and Whitman (1995) and Sondergaard (n.d.), who all confirmed the effectiveness of structure and routine, and state that the quality of life for the individual with PWS depends on the ability of caregivers to provide an environment which is structured enough for the individual concerned. The need to maintain a high level of structure and control in the lives of individuals with PWS puts additional demands on parents and other caregivers.

However, contrary to this, Schwartz et al. (2016) found that the idea that environmental predictability is an important part of managing behaviour problems in individuals with PWS has been challenged, proposing instead that such adherence to routine may lead to more severe outbursts when those routines are subsequently not adhered to. Haig and Woodcock (2017) found that PWS individuals showed less resistance to change if they had been exposed to their parents' or self-imposed flexibility in routines.

- ***Rule-governed behaviour***

Rule-governed behaviour has proven to be effective in managing the undesired behaviour of individuals with PWS. This was achieved by giving clear rules and instructions to be followed and with clear consequences for undesirable behaviour. This is supported by Grant and Evans (1994) who state that rule-governed behaviour is often planned, calculated and logical and it is often said that the person whose behaviour is rule-governed knows the rules. In this study, what made this intervention effective was that it must be implemented with firmness and consistency. The primary caregivers also had clear guidelines they implemented during the outbursts, such as avoiding arguing with the individual during the outburst; remaining calm; sending the individual to his/her room to calm down; and not negotiating with the individual but firmly giving instructions.

- ***Social reinforcers***

Social reinforcers were used by acknowledging and commenting on the PWS individual's positive attributes. Making use of social reinforcers reduced the duration of an outburst when implemented during the episode, and encouraged and empowered individuals to continue with the desired behaviour. This supports the statement that maintenance of positive social contacts with the person, praise, and also stressing the positive aspects of the person with PWS are likely to have a positive effect on the individual concerned (PWSUK, n.d.).

- ***Pivot tool***

The pivot tool, proved to be effective in managing swearing. Swearing in PWS individuals was minimized by ignoring them when they swear and then shifting the focus to other activities. Doing this immobilized the individual and the duration of the episodes was reduced. This is further supported by Whitman and Jackson (2006) who described the praise and ignore (a pivot tool), which illustrates that when inappropriate behaviour occurs the strategy is to pivot attention away from the problem behaviour to another task.

- ***Positive reinforcement***

Positive reinforcements were used, where there was a reward associated with a desired behaviour. This proved to be effective, as it assisted with the improvement in the desired behaviour. This agrees with Corey (2009) who reported that positive reinforcement is often used to increase the frequency of more desirable behaviours, which then replace undesirable behaviours. Some of the effective reinforcements used were rewarding individuals with going to the movies, toys and a "special time" with their favourite person.

- ***Client-centred therapy***

A client-centred approach was adopted with the families, and therefore the individuals with PWS were provided with empathy and a non-judgemental attitude as it became evident that they did not have control of some of the behaviours they were displaying. This approach also allowed the primary caregivers to freely express themselves and share their experiences. It also assisted in reducing the felt stigma regarding their socioeconomic status and their homes.

- ***Grief counselling***

Grief counselling proved to be beneficial for one of the families. It reduced the frequency of outbursts and obsessive-compulsive behaviours of the individual with PWS. This finding suggests that stress associated with grief contributes to the frequency, severity and escalation of some behaviour problems in individuals with PWS. The primary caregiver's grieving process was normalized and she was then able to pay attention to the child and manage his behaviour problems.

- ***Patient participation***

Involving individuals with PWS in decision making also proved to be effective. However, this has to be implemented with great caution. The individual should only be given two options to choose from or two scenarios – to avoid ambiguity and lengthy discussions. This intervention leaves the individual with PWS feeling in control of his/her own treatment plan, encouraged and empowered in the process.

- ***Psycho-education***

Psycho-education was used extensively in this research study and proved to be an effective method. It was used to achieve different goals. First, it was used to teach the families and teachers about the physiological challenges, cognitive impairment and behavioural problems associated with PWS. Secondly, it was used to help families and teachers devise coping strategies and solutions for the problems they were experiencing with PWS individuals. This is affirmed by Ahmed (2004) who highlighted that psycho-education includes teaching coping strategies and problem-solving skills to families, friends, and/or caregivers to help them deal more effectively with the individual. Kodra et al. (2016), in their pilot study, also found that parents of children with PWS need appropriate psycho-education intervention in order to better manage their children with PWS.

Following the psycho-education, the primary caregivers and teachers exhibited empathy towards the individual, and were educated and empowered. Their perceptions regarding the displayed behaviours changed. They became aware that managing and controlling the environment was crucial, and even put structures in place to prevent undesired behaviour from happening. This outcome is similar to the findings of a study of parents of children with intellectual impairment who were

assigned to a psycho-educational group intervention, and who showed significant improvement in parental attitude in relation to child rearing and management of the disability (Lukens & McFarlane, 2004). In addition, the finding concurs with that of Chedd et al. (2006) who stated that the school can benefit from regularly scheduled parent conferences, and also from outside consultation with a professional familiar with PWS.

- ***Psychotherapy for the primary caregiver***

In addition to the stress associated with caring for the individual with PWS, primary caregivers had other personal stressors, such as grief (as explained earlier), marital problems, divorce and unemployment. The stressors influenced their ability to take care of their child with PWS and to manage some of their behaviours – temper tantrums/outbursts and food-related problems. Therefore, psychotherapy was provided to the primary caregivers. It helped to enhance their quality of life and their ability to provide care. This is in line with the findings by Cassidy and Driscoll (2009) and Whittington and Holland (2010) who highlighted that it is important to not only focus on the diagnosed individual but also to provide the necessary intervention to the primary caregivers as they are confronted by multiple stressors which require psychological counselling.

## **9.8 Personal reflections**

Doing the research helped me gain knowledge and a better understanding of PWS, and also the challenges faced by both the diagnosed individual and the primary caregiver. This exploration brought with it an immense learning curve for me as a researcher and as a psychologist.

Doing this research and meeting most of the participants in their homes, was a humbling experience; it is difficult for people to allow strangers into their homes. However, these families opened up their private spaces to me and allowed me to intrude. Not only did they open up their homes but they also allowed themselves to become vulnerable by sharing their experiences and trusting me to assist them. I developed very close relationships with all the research participants.

During the above process it was heart-breaking to see how some of these families were living in poverty. And yet they were inspired to carry on with life. Despite their different socio-economic circumstances, they all had one thing in common: the care and the love the primary caregivers have for their children with PWS, and also the sacrifices they make for them. I was motivated to find ways to assist the research participants to reduce their stress levels. I wish I could have done more for the individuals with PWS but the cognitive impairment was a real stumbling block.

Usually, doing research can be a time-consuming and a daunting process but working with these participants was fulfilling. Not once did I ever feel like giving up. Rather, I was inspired by the experiences of the participants. It was rewarding to implement interventions and to witness them being effective. It was a constant learning process for me.

A challenge for me arose when some of the behaviour problems of the PWS individuals became exaggerated and out of control, and could not be managed by behaviour techniques alone. I felt the need and pressure to refer the individuals concerned to the use of psychotropic medication, in order to control their behaviour. This was not necessarily in the best interests of the individual with PWS but rather was important for the primary caregivers and teachers who were struggling to cope.

I was impressed with the level of care and assistance the families got from government hospitals especially regarding providing participants with GHT, as there are often negative perceptions of government hospitals in the media.

## **9.9 Recommendations**

Based on the findings of this study and all the literature reviewed, it is recommended that the South African government invest more in awareness campaigns particularly for healthcare professionals by conducting workshops on PWS, designing posters and pamphlets with the clinical symptoms and distribute them to clinics and hospitals in order to enable the healthcare professionals to identify PWS early and to manage it effectively. Awareness campaigns are necessary for different communities, both in urban and rural societies.

Genetic counselling should be provided to all parents following the PWS diagnosis, and a clinical psychologist needs to be consulted as soon as the diagnosis is made in order to help the parents and/or primary caregiver cope better with the diagnosis. During genetic counselling, parents should be informed about support systems that are available to them including special schools and the option of a disability grant from government.

Psycho-education on PWS, in a language the primary caregivers understand, should be an integral part of the genetic counselling. Furthermore, this has to be done at different stages of the PWS individual's development. As the individuals with PWS start presenting with food-stealing and other behavioural problems, they should consult with a clinical psychologist to get help with appropriate management. Individual psychotherapy should also be arranged for the parents and/or primary caregivers.

Schools and teachers need to be fully equipped to deal with learners with PWS, and these learners should be placed in a school where the structure is predictable, and with a high teacher-student ratio. It would also benefit families if there was a specialized group home to serve individuals with PWS.

#### **9.10 Limitations and strengths of the research**

One of the families was Afrikaans speaking, and even though they could express themselves in English, there were times when they expressed themselves in Afrikaans – and it was a challenge for me to follow them. Following this experience, I chose research participants who only spoke languages which both I and the participants understood.

The data collection and analysis process were overwhelming and time-consuming. The findings cannot be generalized to the entire PWS population and associated primary caregivers due to the limited number of participants in the study.

Using the language which participants understood helped them to expand more on their experiences, and made them feel more comfortable during interviews and discussions. Working with actual recordings and transcripts was beneficial, as it



allowed me to read through and familiarize myself with the data. The study was qualitative and enabled an in-depth understanding of the experiences of the primary caregivers and diagnosed individuals with PWS.

### **9.11 Suggestions for future research**

Researchers could investigate the experiences of siblings of individuals with PWS.

Future research could be longitudinal in nature, in order to assess the effectiveness of the interventions over a longer period of time. Longitudinal studies will also assist with following these individuals from birth to death, and with reporting on their different experiences through different developmental stages.

Investigating the influence of religion on the ability of the primary caregiver to cope with the diagnosis and challenges of taking care of an individual with PWS, also has potential in terms of possible future research.

Future research needs to look at lived experiences of PWS individuals and their primary caregivers across a number of different provinces in South Africa.

Future research could also understand if there are differing racial and cultural responses in terms of coping with and caring for PWS individuals.

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## APPENDIX I

P.O. BOX 26253  
ARCADIA  
0007  
Cell: 0829572595  
05/03/14

The Prader-Willi Association (South Africa)  
P.O. BOX 2399  
Brooklyn Square  
0075

Dear Sir/ Madam

### **RE: PERMISSION TO CONDUCT RESEARCH WITH YOUR MEMBERS**

I am conducting a study to develop a therapeutic intervention that will be most effective for PWS diagnosed individuals and their primary caregivers. In order to achieve this, it is important to learn about their experiences and challenges. This will be achieved through clinical interviews and implementation of different interventions.

Their individual responses and names will be kept completely confidential. The time frame for participation will depend on the intervention that will be implemented. I am kindly requesting the participation of the Association, the primary caregivers' participation and that of individuals with PWS.

I also hope that the results of the study will share light to psychologists and other health professionals as well as add on the existing literature. I also hope that the results will assist the South African government to provide comprehensive care for the PWS individuals and their families.

Yours Sincerely

Miss. M. Sethuntsa

.....

Clinical Psychologist

## APPENDIX II

P.O. BOX 26253  
ARCADIA  
0007  
Cell: 0829572595  
05/03/14

The Chief Executive Officer  
Dr. George Mukhari Academic Hospital  
P/Bag x 422  
Pretoria  
0001

Dear Sir/ Madam

**RE: PERMISSION TO CONDUCT RESEARCH AT YOUR INSTITUTION**

I am currently in the process of registering with the University of South Africa (UNISA) for Doctoral studies in Psychology. It is a requirement to get permission from the institution where I will conduct my research.

My research topic is entitled: **The development of a therapeutic approach for the treatment of individuals with Prader-Willi Syndrome and their primary caregivers.** I will be conducting interviews and doing psychotherapy with diagnosed Prader-Willi patients and their primary caregivers.

I hereby request your permission to conduct this research at the Dr. George Mukhari Academic Hospital.

Your response will be highly appreciated.

Yours Sincerely

Miss. M. Sethuntsa

.....

Clinical Psychologist

## APPENDIX III

### CONSENT FORM

Title of the study:

**The development of a therapeutic approach for the treatment of individuals with Prader-Willi Syndrome and their primary caregivers.**

.....  
I have been informed about the aims and objectives of the proposed study and was provided the opportunity to ask questions and given adequate time to make an informed decision. The aim and objectives of the study have been explained to me. I have been assured of confidentiality. I have not been pressurized to participate in any way.

I understand that participation in this Clinical Study is completely voluntary and that I may withdraw from it at any time and without supplying reasons. This will have no influence on the regular treatment that holds for my condition neither will it influence the care that I receive from my regular clinician.

I am fully aware that the results of this study will be used for scientific purposes and may be published. I agree to this, provided my privacy and anonymity is guaranteed.

I hereby give consent to participate in this study. I hereby also give permission that the interviews with me and my child/the child I care for, may be recorded for research purposes.

.....  
Name of participant

.....  
Signature of participant

.....  
Name of participant

.....  
Signature of guardian

(NB: For individuals under 18 years)

.....  
Place

.....  
Date



# **APPENDIX IV** **SEMI-STRUCTURED INTERVIEW**

## **1. CONFIDENTIAL BIOGRAPHICAL DATA SHEET: ENGLISH**

<div style="border: 1px solid black; padding: 5px; display: inline-block;">For research purposes</div>	<div style="border: 1px solid black; padding: 5px; display: inline-block;">Research participant code</div>
Date of interview: ..... Time of the interview: .....	

Mark with an X where applicable.

### 1.2 Participant

Caregiver		Individual diagnosed with PWS	
-----------	--	----------------------------------	--

### 1.3 Language of preference

English		IsiXhosa		IsiZulu		Sesotho		SeTswana	
---------	--	----------	--	---------	--	---------	--	----------	--

### 1.4 Gender

Male		Female	
------	--	--------	--

### 1.5 Sexual preference

Heterosexual		Transgender		Gay		Lesbian	Bisexual	
--------------	--	-------------	--	-----	--	---------	----------	--

### 1.5 Race

African		White		Indian		Coloured		Other	
---------	--	-------	--	--------	--	----------	--	-------	--

### 1.6 Age

--

1.7 Place of residence

--

1.8 Mode of transportation

Taxi		Train		Own car		Bus		Other	
------	--	-------	--	---------	--	-----	--	-------	--

1.9 Marital status

1.9.1 Single

No partner (specify reason)		Have a partner		Cohabiting	
-----------------------------	--	----------------	--	------------	--

1.9.2 Married

First marriage		Second marriage		Third marriage or more	
----------------	--	-----------------	--	------------------------	--

1.9.3 Separated

	(specify reason)
--	------------------

1.9.4 Divorced

	(specify reason)
--	------------------

1.9.5 Widowed

--

1.10 Number of dependants

None		1		2		3		4		5 or more	
------	--	---	--	---	--	---	--	---	--	-----------	--

1.11 Occupation (please specify)

--

1.12 Level of education (please specify)

--

### 1.13 Employment

Employed		Part-time		Full-time	
	Unemployed (please provide reason)				

### 1.14 Level of income

None	
Pension fund	
Government Grant	
R1000-R5000	
R6000-R10000	
R11000-R20000	
More than R20000	

### 1. 15 Do you go to church?

Yes		No	
-----	--	----	--

#### (a) If no state reason

--

#### (b) If yes, proceed to answer 1.16 and 1.17

### 1.16 Religion

(specify denomination)
------------------------

### 1.17 Church Attendance

Weekly		Once a month		Once after two to three months		Special ceremony		Once a year	
--------	--	--------------	--	--------------------------------	--	------------------	--	-------------	--

### 1.17 Affiliations .....

## **2. Guidelines to interview questions**

- 2.1 Onset of the diagnosis – age, signs and symptoms
- 2.2 Impact of the diagnosis on the individual and family
- 2.3 Experience of taking care of a PWS individual
- 2.4 Challenges of living with PWS / PWS diagnosed individual
- 2.5. Dietary challenges and food storage
- 2.5 Types of treatments
- 2.6 Health care professionals
- 2.7 Adherence to treatment
- 2.7 Relationship with the PWS diagnosed individual / relationship with the caregiver
- 2.8 Support systems
- 2.10 Behaviour changes and interventions
- 2.11 Psycho-education on future physical, behavioural, emotional, and cognitive changes
- 2.12 Needs of the caregiver / needs of the PWS diagnosed individual
- 2.16. Leisure activities / hobbies
- 2.18. Future plans
- 2.19. Additional information
- 2.20 Recommendations